VILNIUS UNIVERSITY
FACULTY OF MEDICINE

Integrated Studies of Medicine

Institute of Clinical medicine, Clinic of Gastroenterology, Nephro-Urology and Surgery

Hans Schröder, Year 6, Group 9

INTEGRATED STUDY MASTER'S THESIS

Congenital Scoliosis

Supervisor: Assist. Prof. Dr. Giedrius Bernotavičius

Head of the department: Prof. Dr. Habil. Kestutis Strupas

Vilnius, 2025

hans.schroeder@mf.stud.vu.lt

Abbreviations

AI – Artificial Intelligence

CA – Cobb Angle

CT – Computed Tomography

CVM – Congenital Vertebral Malformations

MRI – Magnetic Resonance Imaging

PSO – Pedicle Subtraction Osteotomy

RVAD – Rib-Vertebral Angle Difference

SCD – Spondylocostal Dysostosis

TIS – Thoracic Insufficiency Syndrome

VACTERL – Vertebral defects, anal atresia, cardiac defects, trachea-esophageal fistula, renal anomalies, and limb abnormalities

VEPTR – Vertical Expandable Prosthetic Rib

3D – Three dimensional

List of figures:

Outline

1. Summary

Congenital scoliosis is a complex spinal deformity caused by a malformation of the vertebral bodies during embryonic development. It is often associated with other systemic anomalies. The prognosis depends largely on the type, location and number of malformations, as well as any associated intra-, or extraspinal pathologies. Early diagnosis is crucial, as the first years of life are considered to be a critical period for the course of the disease and can be decisive for the success of treatment. The therapeutic approach must be individually adapted to the individual child and be multidisciplinary in nature, this is the only way to take account of both structural features and functional needs. Growth-directed procedures such as convex instrumented hemiepiphyseodesis with concave distraction allow correction of the deformity while preserving thoracic and spinal growth. In the case of a 13-month-old child with multiple deformities and concomitant syringomyelia, the need for staged surgical intervention, including shunt implantation, hemivertebrectomy and pedicle subtraction osteotomy (PSO), was demonstrated to both secure neurological function and achieve structural correction. Complications such as implant failure highlight the importance of continuous imaging and a dynamic therapeutic approach. Forward-looking technologies such as AI-based image analysis, patient-specific 3D models and epigenetic research promise to further improve treatment planning and prognosis. Early detection and precise, personalised treatment are essential to avoid serious long-term consequences such as thoracic insufficiency syndrome (TIS) and to ensure long-term quality of life for affected children.

2. Keywords

Congenital scoliosis, vertebral malformations, hemivertebra, kyphoscoliosis, growth modulation, thoracic insufficiency syndrome, convex hemiepiphysiodesis with concave distraction, imaging, epigenetic, multimodal imaging, AI

3. Methodology

For the preparation of this paper, systemic literature research was conducted via PubMed, Google Scholar, and specialist databases and focused on publications from 2015 to 2024 in order to reflect the current state of scientific discussion, clinical diagnostics, and surgical treatment options. Systemic reviews, guidelines, case series, as well as original papers related to etiology, classification, imaging, and treatment of congenital scoliosis were considered. In addition, the Orthobullets platform was used. The purpose of this review is to present a structured overview of current clinical guidelines and treatment options for congenital scoliosis, incorporating a clinical case to contextualize the provided

theoretical information. Although the methodological focus was on current literature, the work of Mehta (1972) was explicitly included in the analysis as it represents a significant historical milestone in the diagnosis of infantile scoliosis. For practical illustration, a clinical case of a 13-month-old boy with multiple congenital malformations was analyzed retrospectively. The clinical, radiological, and surgical findings were reconstructed on the basis of existing documentation and compared with findings from the literature. The selection and evaluation of surgical interventions were based on recognized decision-making aids and individual anatomical conditions.

In addition, promising technological developments were critically reflected upon in the discussion to highlight potential perspectives for improved care.

4. Introduction

Congenital scoliosis refers to a complex and heterogeneous group of spinal deformities that originate in the incorrect embryonic segmentation or formation of the vertebral bodies between the 4th and 6th week of gestation (1,2). The prevalence is between 0,5 and 1 per 1000 live births, with many cases only being diagnosed late by incidental findings, which indicates a high number of unreported cases (2). Overall congenital scoliosis accounts for around 10% of all scoliosis cases, with the course of the disease varying greatly from individual to individual: while a quarter of deformities are non-progressive, around 50% are progressive, sometimes with a significant impact on body posture and organ function (2). The pathogenesis is multifactorial with genetic factors, such as mutations in the TBX6 or FBN1 gene, and environmental factors, such as maternal diabetes or alcohol exposure (2).

In many cases, congenital scoliosis is not limited to the spine. Frequently, patients have additional abnormalities of other organ systems, particularly the cardiovascular, urogenital, or central nervous system (3).

Prognosis of congenital scoliosis is not only dependent on the type of vertebral malformation, but also on its location, number and possible combinations (1,4). Early assessment of prognosis is particularly important in the first few years of life, which is considered to be a critical period for future outcomes (2)

Clinically, the care of these children requires a high degree of individualized treatment. The course of the disease is often difficult to predict and is characterized by recurrent diagnostic and therapeutic decisions which aim to prevent later complications (2).

In the context of these complex medical and surgical challenges, this work aims to provide a structured review of current concepts on the pathogenesis, diagnosis, and treatment of congenital scoliosis. The focus is on a clinical case of a 13-month-old boy with multiple spinal deformities who underwent a growth-directing, multi-stage treatment with accompanying complications. The purpose

of this particular case analysis is to illustrate the complex reality of the care of affected children and underline the need for individualized treatment paths.
The conclusion will evaluate the selected operative method used in this case in comparison with other possible options.
In addition, the concluding discussion will reflect promising future developments such as epigenetic approaches, AI-supported image analyses, and the use of patient-specific 3D models to highlight possible perspectives for improving patient care.

5. Literature review

5.1. Etiology

The exact etiology of congenital scoliosis is not entirely understood, but it is likely multifactorial, including environmental and genetic factors (2).

5.1.1. Embryological development and pathogenesis

To truly understand congenital scoliosis, it is important to understand the embryonic development of the human body. This is because it is at this stage that fundamental processes are started that are crucial for the development, classification, and progression of the disease. Therefore, the following section will take a closer look at the development of the spine during embryogenesis and its significance for the pathogenesis of congenital scoliosis. The third week of embryonic development is particularly important. During this phase, the bilaminar embryonic disc develops into the trilaminar disc, consisting of ectoderm, mesoderm and endoderm.
This process is known as gastrulation. Among other things, the notochord develops from the mesoderm and serves as an important signalling structure. It triggers the formation of the neural tube from the ectoderm, which later becomes the central nervous system. At the same time, the paraxial mesoderm differentiates into somites. These are the precursors of the vertebral bodies, ribs and associated muscles. Disruptions in this developmental process can lead to malformations of the spine, and thus to the development of congenital scoliosis. Segmentation of the somites occurs in a head-to-tail sequence and is regulated by signaling pathways, including FGF, WNT, Notch, and retinoic acid gradients. In the fifth week of gestation, the somites differentiate into dermatome, sclerotome, and myotome, and the cells of paired sclerotomes migrate to surround the notochord and neural tube, forming the precursors of the vertebrae. The intervertebral discs arise from a cell-free space between the neighboring sclerotomes, while the vertebral bodies develop by fusion of the rostral and caudal sclerotome domains. During the sixth week, the developing vertebrae begin the process of chondrification, the transition from mesenchymal tissue to cartilage. A disruption in this mechanism

or in one of the tightly controlled pathways can result in vertebral malformations contributing to congenital scoliosis (1,2,5).

5.1.2. Environmental factors

There are different environmental factors that are thought to have a positive relationship with congenital vertebral malformations (CVM). One of the most significant environmental influences in congenital scoliosis is fetal hypoxia, which can arise from maternal smoking, high altitude exposure, or carbon monoxide exposure. It was shown in a mouse model that in an acute carbon monoxide exposure at 9.5 days of gestation of mice, which correlates with the thoracolumbar spine development in humans, there was a high degree of vertebral abnormalities. Another study demonstrated that hypobaric hypoxia on day 10 of gestation in mice resulted in vertebral defects (6).

Short-term oxygen deprivation has been found to result in a secondary downstream effect on the Notch pathway. As was mentioned earlier, it is a key regulator in somite formation and vertebral segmentation. Interference with this signaling pathway can result in somite compartmentalization errors and vertebral malformations (6). It has been shown that hypoxia promotes the development of reactive oxygen species, leading to oxidative stress and cellular damage (5) . This imbalance promotes the apoptotic process and decreases the expression of Pax3, a gene that plays a central role in somite formation and neural tube closure (5,7). Additionally, early hypoxic conditions are shown to influence the vascular development of intersegmental arteries, which are important for resegmentation and early chondrification of the vertebral bodies (6).

Maternal diabetes is another factor contributing to the development of congenital scoliosis. It is shown that hyperglycemia induces an increased production of reactive oxygen species, which causes apoptosis in the developing somites (1,2,5). Other teratogenic factors include hyperthermia, whereby proteins in the notch pathway are bound with heat shock proteins. Reactive oxygen species also contribute to the teratogenic effects of maternal smoking and valproic acid exposure during pregnancy. Maternal smoking is a source of carbon monoxide, which leads to reactive oxygen species production (1,2,5).

There are no endocrine factors that have been considered as a possible cause of congenital scoliosis (2).

5.2. Genetic factors

Genetic factors in the development of congenital scoliosis remain largely unclear, exome sequencing identified potentially associated genes. Studies indicate that a combination of a TBX6 null mutation and a hypomorphic allele may account for approximately 11% of sporadic congenital scoliosis cases (8). TBX6 is essential in the process of somitogenesis (9). The Notch signaling pathway genes,

including DLL3, MESP2, LFNG, HES7, and RIPPLY2, are also associated with somitogenesis, and their mutations have been identified in spondylocostal dysostosis (SCD), but no definitive causative link to congenital scoliosis has been established (10). Although mutations in the LFNG gene are primarily associated with SCD, they also have been reported in isolated cases of patients with congenital scoliosis (10). The FBN1 gene is coding for fibrillin-1, a glycoprotein which is essential for the formation of microfibrils in connective tissue. Mutations in this gene are known for their association with Marfan syndrome, but they have also been proposed as a possible monogenic cause of congenital scoliosis (2). Marfan syndrome and other connective tissue disorders such as Beals, congenital muscular dystrophies, and hypotonia are also associated with congenital scoliosis (2).

5.3. Physical examination and history taking

The diagnosis of congenital scoliosis is often made incidentally, for example during physical examinations or imaging studies for other medical reasons (5). Malformations can usually be detected prenatally by ultrasound between 20 and 28 weeks of pregnancy, and in some cases as early as 12 weeks. Despite this, 25% of affected infants receive a diagnosis within the first year, and nearly 50% of cases are confirmed by the age of three (5). The diagnosis of congenital scoliosis is made through clinical evaluation, imaging studies, and genetic testing. A comprehensive family medical history, review of obstetric records and antenatal imaging are essential for the prenatal detection of possible spinal malformations and the planning of an individualized treatment plan (2,11). A careful medical history is vital to identify known risk factors such as smoking, diabetes, or substance use (2). During the clinical examination, abnormalities in the coronal, sagittal, trunk and pelvic alignment should be carefully documented (5,12). It is also important to note the child's age, height and weight, as these parameters are important predictors of skeletal growth and the likelihood of progression of the curvature (5). Thoracic insufficiency syndrome, which refers to the inability of the thorax to support normal respiration and lung growth, can result from compression of the height and depth of the rib cage as a consequence of congenital scoliosis (5,13). The thumb excursion test should be performed to assess lung function, and the baby should be examined for deformities, asymmetry, and the excursion and inspiratory/expiratory function of the chest wall. Furthermore, the neurological status should be evaluated, and the body should be checked for skin and orthopedic anomalies (5).

The malformations of congenital scoliosis are not limited to vertebral malformations but are often multisystemic, with skeletal and non-skeletal malformations beyond the vertebral malformations in 30 to 60% of cases. Congenital scoliosis is commonly associated with syndromes such as VACTERL, Goldenhar syndrome, Klippel-Feil syndrome, and Jarcho-Levin syndrome (1–3,5).

The reported percentages of anomalies in congenital scoliosis vary significantly across different studies. Cardiac anomalies have been reported in 4.9% to 54% of congenital scoliosis patients, with ventricular septal defect being one of the most common cardiac anomalies (4,5,11). Moreover, urogenital anomalies range from 4.9% to 40%, and intraspinal anomalies vary from 10% to 58%, both among the most common anomalies in patients with congenital scoliosis (1,3,5). When one nonskeletal abnormality is discovered, it predicts the presence of other abnormalities, and the location of the curve may indicate the associated abnormality (5).

5.4. Imaging

A combination of imaging studies is essential for the accurate diagnosis and treatment plan of congenital scoliosis. They are used to assess vertebral anomalies, curve progression as well as associated anomalies. Anteroposterior and lateral plain X-ray imaging is the first-line diagnostic tool (2). Measurement of the Cobb angle (CA) is a useful method for quantitative assessment of spinal curvature (14). It can also be used to determine the extent of segmentation and formation defects and to monitor disease progression over time, making it a central component of routine follow-up in scoliosis (3,5). Computed Tomography (CT) scans provide a more detailed visualization of the vertebral anomalies, allowing a better understanding of the bony structures and deformities. This makes them in particular useful in pre-operative planning and complex cases (11). Since the radiation exposure is increased compared to other imaging modalities, it should be limited to cases where you need additional anatomical details especially in preoperative planning(5). It can furthermore be used to detect if thoracic insufficiency syndrome is present (2). Magnetic resonance imaging (MRI) is indicated in all patients with congenital scoliosis, particularly before surgery, to detect spinal cord malformations such as tethered cord, syringomyelia, diastematomyelia, or cysts (1–3,5,15). Ultrasonography can detect vertebral anomalies as early as the second trimester, making it a valuable imaging tool for diagnosis (11). It is effective in identifying structural abnormalities but has limited sensitivity in detecting minor vertebral defects, often necessitating follow- up postnatal imaging (5). Since it is radiation- free and non- invasive, ultrasonography can assess related organ system anomalies. Given that urogenital anomalies occur in 4. 9% to 40% of patients with congenital scoliosis, a renal ultrasound plays a crucial role and should be performed during postnatal assessment (3). Furthermore, echocardiography should be conducted, considering the frequent association between congenital scoliosis and congenital heart defects (3,16). In summary, each imaging modality contributes to a comprehensive evaluation of patients with congenital scoliosis. X- rays are essential for initial assessment and monitoring progression; MRI is important for detecting intraspinal

anomalies; CT scans provide critical visualization of bony structures, especially before surgery; and ultrasound aids in the detection and assessment of associated anomalies.

5.5. Classification

Accurate classification is essential for understanding the nature of the deformity, predicting its progression, and selecting appropriate treatment approaches (17). Over time, classification systems have evolved from simple radiographic observations to advanced three-dimensional image-based models.

The classification method proposed by Winter and collaborators in 1968, shown in figure 1, was the earliest and was based on radiographic findings (17).

This classification organizes congenital scoliosis into three primary categories: failures of formation, failures of segmentation, and mixed anomalies. Failures of formation occur when a vertebra does not fully develop, leading to asymmetrical spinal growth and scoliosis (18). They can be subdivided into semisegmented hemivertebrae, fully segmented hemivertebrae, and wedge vertebrae. A fully segmented hemivertebra has normal intervertebral discs above and below, allowing for independent growth (18). It has a higher potential for progression, particularly if it is unilateral, as it promotes asymmetrical growth (18). The partially segmented hemivertebra (semisegmented in Figure 1) is fused to an adjacent vertebra on one side while maintaining an open disc space on the other side, which limits its growth potential compared to the fully segmented hemivertebra (18). Another hemivertebra that forms as a result of a failure of formation is the wedge vertebra. This type represents a partial failure of vertebral body formation, where one side of the vertebra is underdeveloped, leading to an angular deformity (18). Failure of segmentation occurs if two or more vertebrae fail to separate properly, which leads to rigid spinal segments that restrict normal growth. They can be subdivided into block vertebrae and unsegmented vertebrae. Block vertebrae, which are the least likely to cause any significant deformity, result from bilateral segmentation defects where adjacent vertebrae are completely fused, eliminating normal disc spaces and preventing movement at the affected part (18). A unilateral unsegmented bar is a bony bar on one side of the spine, often on the concave side of the scoliosis curve, which fuses both the disc spaces and/or the facets and does not grow (18). Mixed failures involve a combination of formation and segmentation defects. The hemivertebra with a contralateral unsegmented bar has the highest potential for progression and represents a condition where a hemivertebra on one side causes asymmetric growth, while an unsegmented bar on the opposite side restricts movement, leading to a severe curve (18).

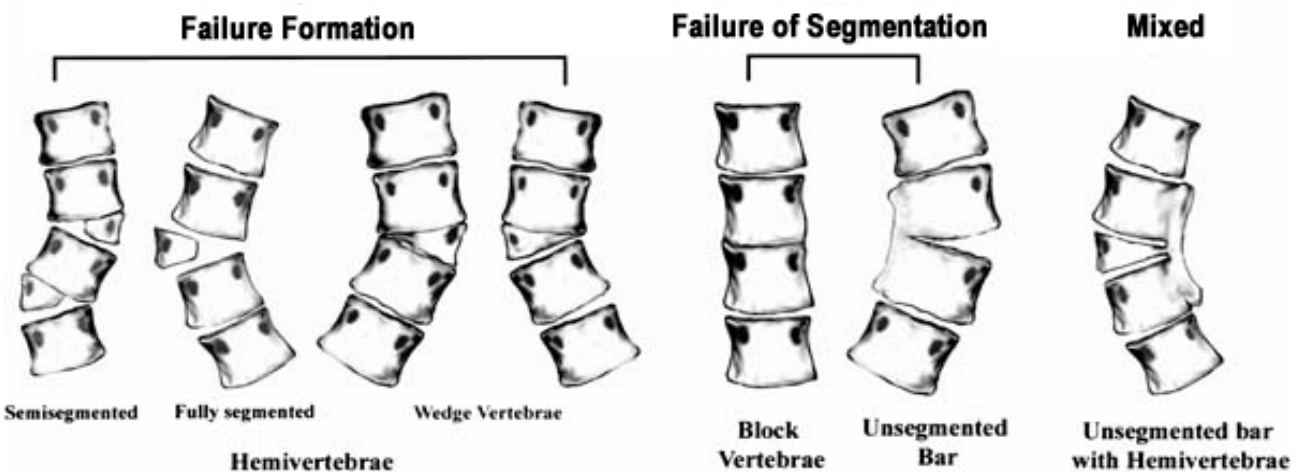

Figure 1: Reference: 1

With the advancement in medical imaging, new classification models emerged. In 2004, Imagama and colleagues introduced a system that used three-dimensional imaging to analyze vertebral anomalies more precisely. In addition to the formation errors, they included the structural components of the anterior and posterior vertebrae (17).

In 2009, Kawakami expanded upon previous models by incorporating three-dimensional computed tomography imaging. This classification, shown in figure 2, differentiates between solitary simple malformations, multiple simple anomalies, complex anomalies, and pure segmentation failures (17).

Type 1	Solitary simple (unison) type
	Hemivertebra
	Wedge vertebra
	Butterfly vertebra
	Defect
	Others
Type 2	Multiple simple (unison) type
	Combination of hemivertebra, wedge vertebra, or butterfly vertebra
	Discrete, adjacent, or others
Type 3	Complex (discordant) type
	Mismatched complex type
	Mixed complex type
Type 4	No abnormal formation type
	Pure segmentation failure

Figure 2: Reference 50

In 2015, Burnei and Gavriliu proposed a more recent classification, shown in figure 3. They consider not only vertebral formation and segmentation defects but also the role of longitudinal and rotational imbalances in the progression of scoliosis. In this classification, a distinction is made between scoliosis due to longitudinal imbalance, which is caused by vertebral malformations that primarily

affect the coronal plane, and scoliosis due to rotational imbalance, which is affected by asymmetric vertebral growth and unequal distribution of forces along the spine (17). Approximately 80% of congenital scoliosis cases are due to abnormalities in vertebral formation or segmentation, with the remaining 20% being a combination of both abnormalities (18).

Classification of congenital scoliosis has evolved considerably over time. Whereas early systems, such as the Winter classification, were primarily based on radiographic criteria, modern approaches, such as the Burnei-Gavriliu classification, offer a more sophisticated and predictive model for diagnosis and treatment planning of spinal deformities (17).

Treatment of congenital scoliosis is based on several factors, including the number of affected vertebral segments, the degree of curvature, the presence of fused ribs, and the skeletal maturity of the patient.(17). Approximately 80% of congenital scoliosis cases are either formation or segmentation failures, while the remaining 20% consist of mixed anomalies (18). The classification of congenital scoliosis has evolved significantly. While early systems, such as Winter´s, relied primarily on radiographic assessments, contemporary methods, including the Burnei-Gavriliu classification, offer a more detailed and predictive framework for the diagnosis and management of spinal deformities (17).

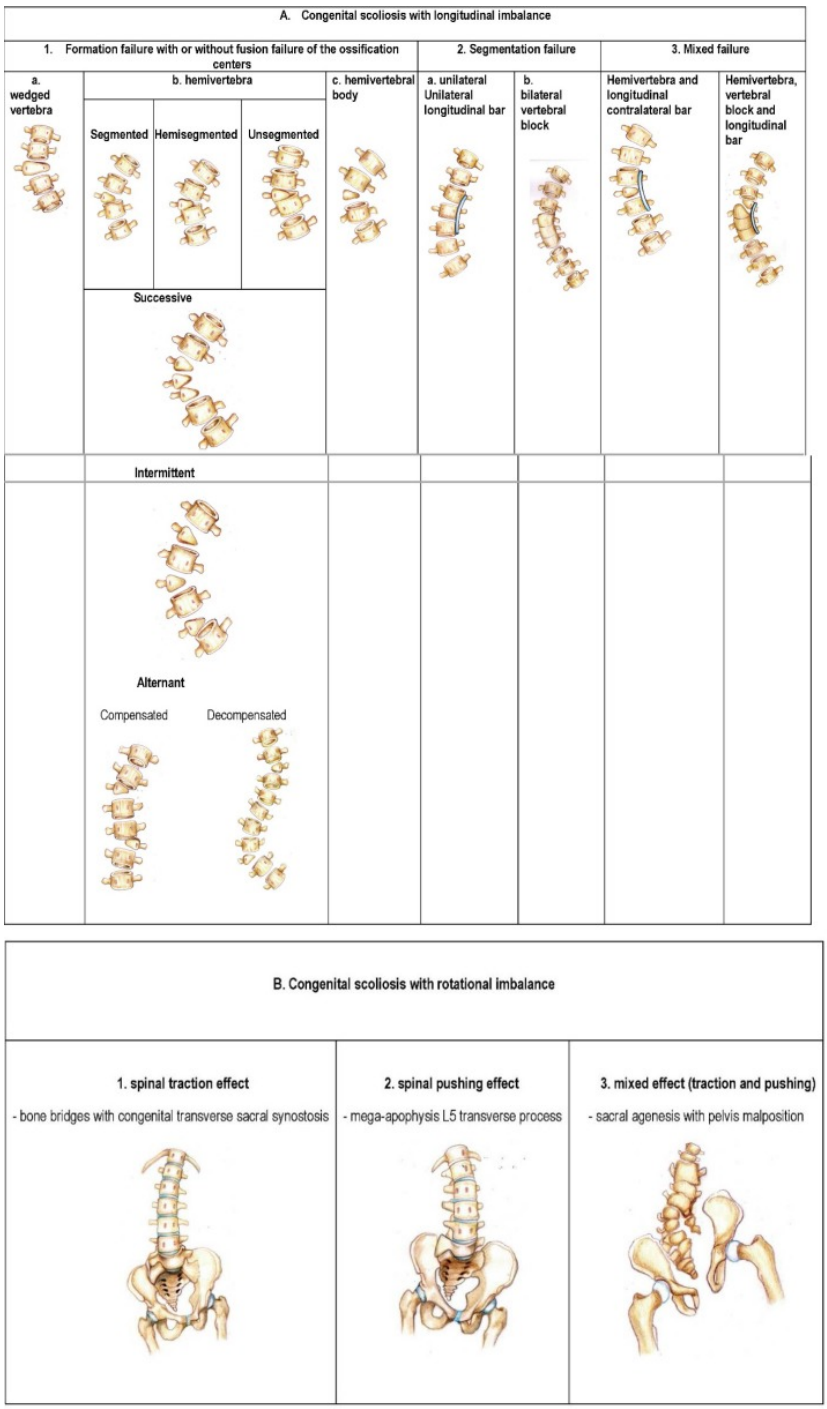

Figure 3: Reference 13

5.6. Treatment

The management of congenital scoliosis depends on several factors, including the number of affected vertebral segments, the severity of the curve, the presence of fused ribs, and the patient's skeletal maturity. Since congenital scoliosis arises from structural vertebral anomalies, non-surgical approaches have limited effectiveness; thus, surgical intervention is often necessary to prevent curve progression. A structured treatment approach can be visualized using the guide to surgical decision-making proposed by Agnivesh Tikoo and colleagues, illustrated in figure 4.

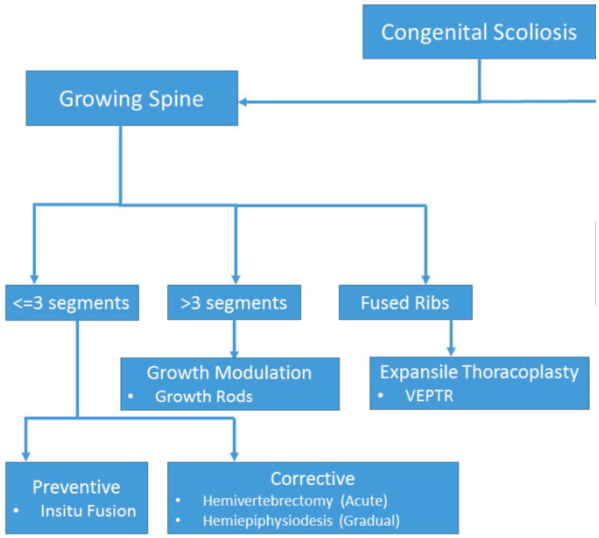

Figure 4: Reference 16

5.6.1. Non-surgical treatment

Non-surgical treatment approaches include the use of bracing and close monitoring (1). Bracing is commonly used in idiopathic scoliosis to arrest the curve progression during growth (19). However, its effectiveness in congenital scoliosis is reduced due to the structural rigidity of the vertebral anomalies (1,2,18). Observation is favored in patients with mild, non-progressive congenital spinal deformities, like incarcerated hemivertebrae, nonsegmental hemivertebrae, and some partially segmented hemivertebrae. This includes regular radiographic and clinical monitoring without immediate intervention (1,20).

5.6.2. Surgical treatment

In situ fusion involves stabilizing the vertebrae in their current alignment without significant deformity correction. It primarily addresses progressive, minimal deformities associated with unilateral unsegmented bars (1). Typically, an improvement in curvature of about 10° to 15° can be achieved with instrumental correction (2). Possible complications include pseudarthrosis, loss of correction and the crankshaft phenomenon, which can occur particularly in younger patients (2). Anterior diskectomy has been shown to enhance stability and prevent the crankshaft phenomenon (2). Since a multi-level arthrodesis is known to decrease the vital capacity, it is recommended to extend along a short segment (2). It is recommended for younger patients with progressive non-deforming curves not exceeding 40° angles (2).

Hemivertebrectomy is a surgical procedure where the malformed vertebra, along with the neighboring disks, laminae, and pedicles, is excised to rectify severe spinal imbalance (1,2). With reported

corrections up to 40°, it provides better curve correction outcomes compared to in-situ fusion and hemiepiphysiodesis (2). This technique is recommended in the case of patients under six years with hemivertebrae, especially in the thoracolumbar and lumbar spinal regions, which are less vulnerable to neurological damage during manipulation, with a curve exceeding 40° (1,2). Hemiepiphysiodesis is a surgical procedure that deliberately stops the growth of the vertebral bodies on the convex side of the curve. At the same time, the concave side is allowed to continue growing, gradually correcting the curvature over time (2). This method is suited for patients younger than five years, presenting with fully segmented hemivertebrae and a moderate curvature of less than 70° without significant kyphosis and a short segment of vertebrae (2).

Growing rods is a technique used for long-segment progressive deformities in young patients with the goal of controlling the spinal curvature but still allowing progressive spinal elongation to maintain pulmonary function and avoid early fusion (2,5,21). The rods are anchored above and below the curve and undergo lengthening every four to six months until final fusion is performed (2,5). The dual-rod technique, which has shown a Cobb angle improvement of 29-50%, is superior in terms of stability and correction compared to the single-rod approach (2,5). Disadvantages are the frequent procedures, the increased radiation due to repeated imaging, and the chance of autofusion due to the repeated lengthening procedures (2,5). Complications, including implant-related issues such as rod breakage and anchor failure, occur in approximately 50% of cases with growing rods (5,22).

Expansile thoracoplasty is a surgical approach aimed at enlarging the thoracic cavity and improving respiratory function (5). It is indicated in congenital scoliosis patients with multiple rib fusion or absence, whereby thoracic insufficiency syndrome can develop (2). The Vertical Expandable Prothetic Titanium Rib (VEPTR) is a primary device used in the expansile thoracoplasty which is hooked around the ribs, spine, or pelvis, depending on the severity of the deformity and is the only effective solution in the surgical treatment of TIS (2,23). Its lengthening occurs every five to six months on the concave side of the curve (2,5).

Vertebral Column Resection (VCR) is a surgical procedure used in cases with ridgid residual curves, neurological deficits or failure of previous treatments. It involves the complete removal of vertebral segments, including the vertebral body, lamina, transverse processes and, if necessary, parts of the ribs. The spine is then stabilized to ensure correction and restore stability (2). The primary objective of this reconstructive osteotomy is to restore spinal balance and remove fusion masses, but it is associated with neurological complications in 8% of cases and spinal injury in 2% of patients (2,24).

Figure 5 presents a treatment algorithm proposed by Armer Sebaaly and colleagues, whose main criteria for determining an optimal treatment plan include age, the magnitude of the Cobb angle, and the type of formation (2).

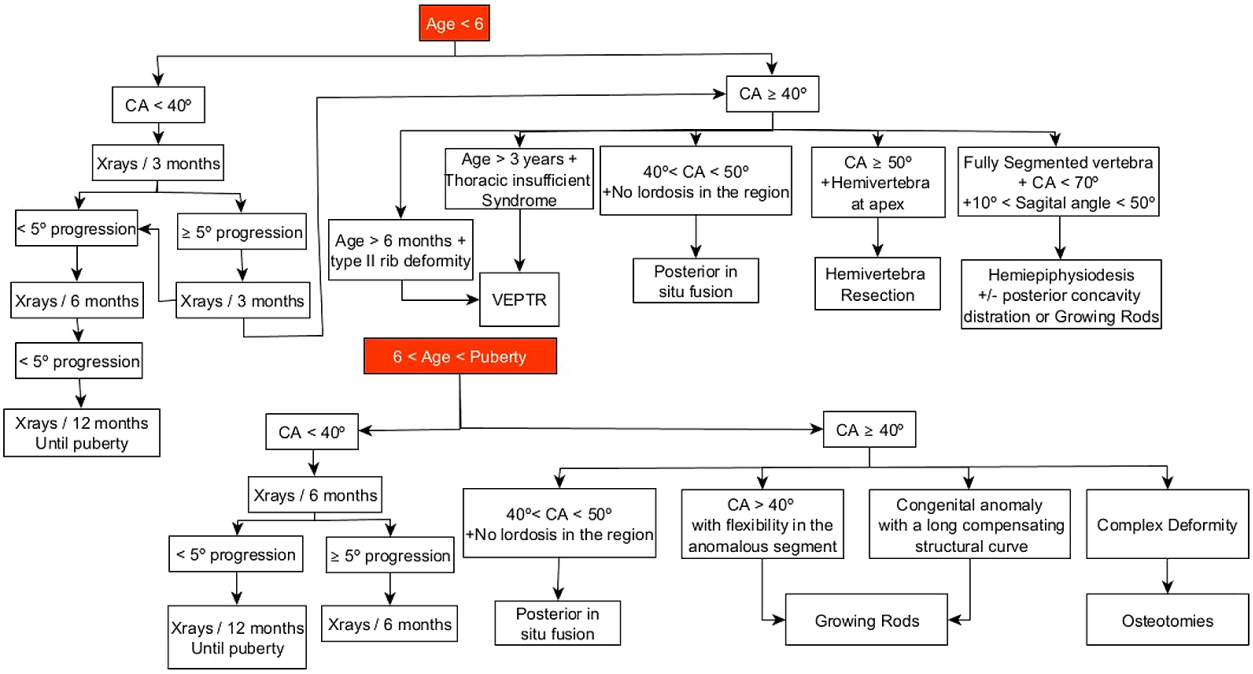

Figure 5: Reference 2

5.7. Impact of congenital scoliosis on the Quality of life

The occurrence of non-spinal anomalies, particularly cardiac and renal malformations, is very common in patients with congenital scoliosis (CS). These comorbidities can lead to increased health burden, limited physical activity, and reduced quality of life, highlighting the need for a multidisciplinary approach to the management of these patients.

Children with congenital scoliosis who received a Vertical Expandable Prosthetic Titanium Rib had better functional outcomes and body image satisfaction compared with patients who received conservative treatment or spinal fusion (18). These results suggest that early treatment with expandable implants preserves growth potential, improves lung function and contributes to a better quality of life in the long term. In particular, children with TIS have a lower quality of life compared to patients with chronic heart disease or malignancy, highlighting the importance of prevention and early intervention (13).

In addition to physical limitations, congenital scoliosis can be psychologically distressing. The visible deformity and associated functional limitations can lead to psychological distress, low self-esteem and a significantly reduced overall quality of life (25). In addition, studies show that affected children have an increased risk of anxiety and depression (26).

6. Case presentation

Congenital scoliosis is a challenging deformity resulting from vertebral anomalies that occur during embryonic development. In this chapter, we will discuss a complex case of a 13-month-old boy with multiple congenital spinal anomalies, progressive kyphoscoliosis, and intraspinal pathology. This case illustrates the challenges a spine surgeon has to face and also the surgical management of congenital scoliosis. Furthermore, it emphasizes the possible complications like rod breakage, implant dislocation, and emerging vertebral anomalies at different levels. Moreover, it highlights key principles, which include an early diagnosis, the need for repeated interventions, and the need for a detailed investigation.

Our patient is a 13-month-old boy who presented with a noticeable spinal deformity. No records regarding prenatal complications, family history, or maternal risk factors were available. On initial evaluation, his neurological exam showed no motor or sensory deficit. His physical appearance was remarkable for significant kyphoscoliosis. No information about cutaneous markers or limb deformities were available.

Figure 6 shows a 3D CT reconstruction of the patient's thoracic and lumbar spine. A thoracic scoliosis with a curvature to the left and a lumbar curvature to the right are visible. Vertebral anomalies such as a hemivertebra, block vertebra, and wedge vertebra are present. Asymmetric rib arches indicate rotational deformity.

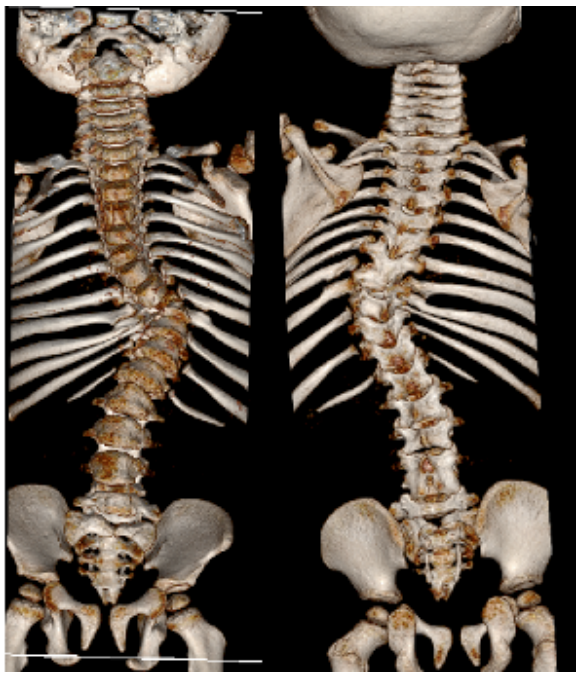

Figure 6: 3D – reconstructed CT scan of the thoracic and lumbar spine showing a left -sided thoracic scoliosis and right sided lumbar curve. Multiple vertebral anomalies are visible, including hemivertebreae, block vertebrae, and wedge deformities.

One of the key metrics for evaluating the severity of congenital scoliosis and estimating the potential for curve progression is the Cobb angle, which remains the gold standard when measured on radiographic films (2,27). To determine the Cobb angle, the Cobb- Lippmann technique is used. Lines are drawn along the upper endplate of the uppermost vertebrae and the lower endplate of the lowest vertebra. The angle of interest is the one formed by the intersection of these lines (2). Scoliosis is a Cobb angle ≥ 10°, and a Cobb angle < 10° is called spinal asymmetry (28). The concept of the rib-vertebral angle difference, or RVAD, was originally introduced in the early 1970s by Dr. Mehta to differentiate progressive from resolving infantile scoliosis. The rib-vertebral angle is calculated by examining both the concave and convex sides of the apical thoracic vertebra, where each side´s angle is formed by the vertebra and its corresponding rib. The R-V angle difference is the disparity between the angles on these two sides (29). Mehta observed that 20 degrees serves as a critical cutoff. In cases infantile scoliosis cases that later resolved spontaneously, the initial R-V angle difference was typically under 20°. Conversely, curves that eventually progressed often began with an R-V angle difference of at least 20° (29). In our case, the X-ray of the 13-month-old boy, presented in figure 7, showed a Cobb angle of 52.7° and an RVAD of 41°, which makes a progression very likely.

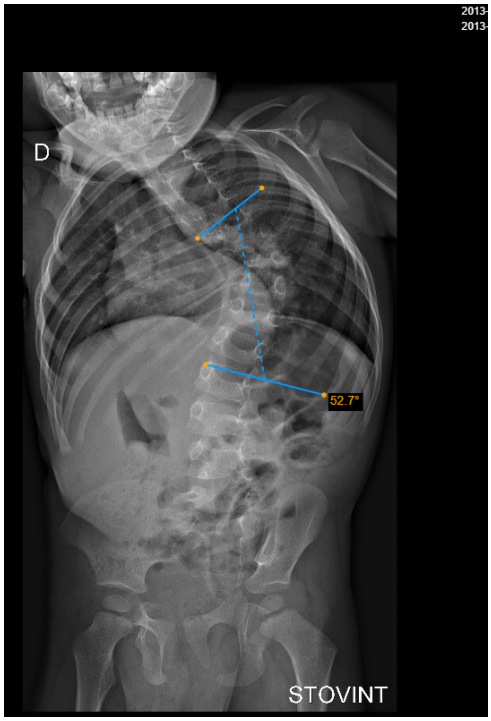

Figure 7: Upright AP radiograph of the patients spine showing a primary thoracic scoliosis with a Cobb angle of 52.7°.

The lateral radiograph of our patient, shown in figure 8, revealed a pronounced thoracic kyphosis of 73.2°, surpassing the 20° cutoff typically used to define a pathologic curve in young children with spinal deformities (30).

Congenital kyphosis, particularly cases measuring well above 45°, typically necessitates surgical rather than conservative treatment to restore sagittal balance and reduce the risk of neurological compromise (31).

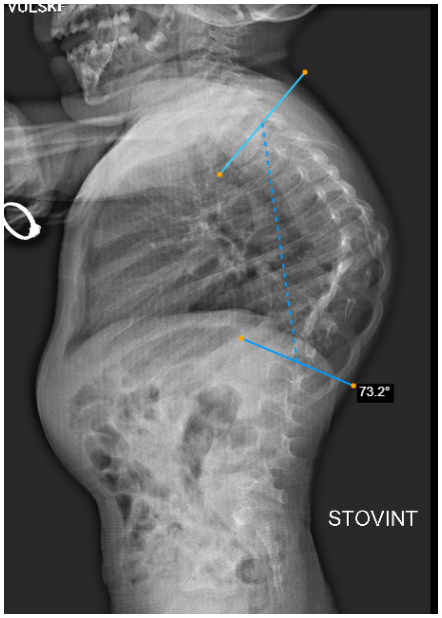

Figure 8: Lateral radiohgraph showing a severe kyphotic deformity with a measured kyphosis angle of 73.2°.

As mentioned, MRI imaging should be done to exclude spinal abnormalities prior to surgery. In this case, an MRI revealed a syrinx from Th2-Th3 to Th12-L1 disc level.
Syringomyelia is with 5 to 20.2% quite common in patients with congenital scoliosis (2,32). The presence of a syrinx can exacerbate a spinal deformity, triggering or worsening scoliosis or kyphosis in pediatric patients (33). An early detection via MRI is essential because its identification and treatment can reverse the deformity (33). Furthermore, the MRI of our patient showed a significant kyphoscoliosis of the thoracic spine to the left and lumbar to the right.
Moreover, it revealed signs of spinal canal stenosis at Th8 and L4 levels.

In this patient, the preoperative MRI confirmed a large syrinx extending from T2-T3 down to T12-L1, which posed a risk of neurological complications (Figure 9,10). As a result, the surgical team decided to perform a syringo-subarachnoid-peritoneal shunt to decompress the intramedullary cavity before addressing the spinal anomaly. The aim was to stabilize the intramedullary pressure to minimize the risk of acute spinal cord injury during subsequent surgery. In this case, the team utilized a syringo-subarachnoid-peritoneal route to divert syrinx fluid into the peritoneal cavity alleviating cord expansion. This measure offered the best immediate safeguard against neurological deficits. It decreases the risk of neurological compromise during correction surgeries (34).

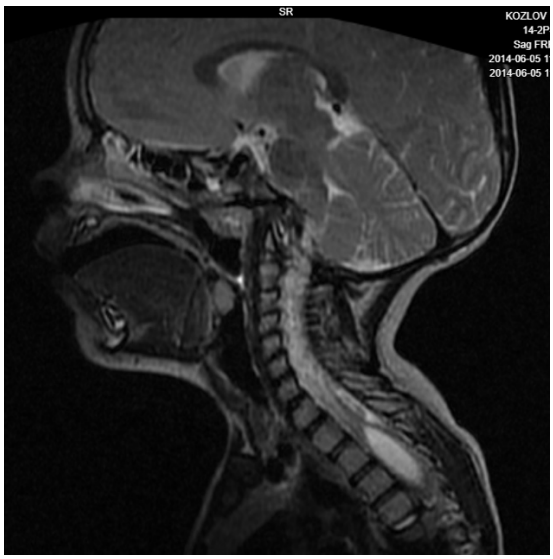

Figure 9: Sagittal T2-weighted MRI of the cervical and thoracic spine showing a large intramedullary syrinx.

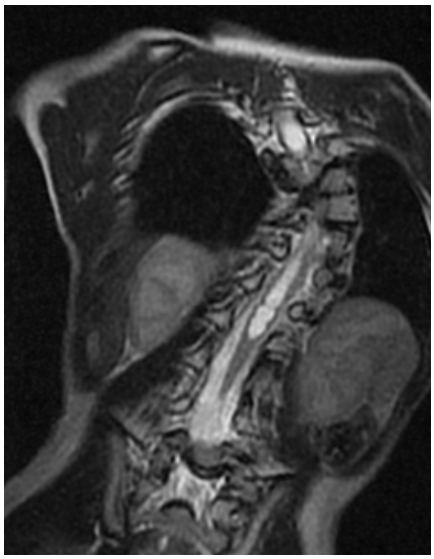

Figure 10: Coronal T2- weighted MRI revealing a syrinx extending across multiple segments.

Despite the progression, the child was not yet demonstrating accelerated progression or neurological compromise. Therefore, the initial plan was to observe. Regular follow-up X-rays were arranged to monitor the angles. The decision for the surgery with the best outcome is very complex in most cases of congenital scoliosis. (Figure 12).

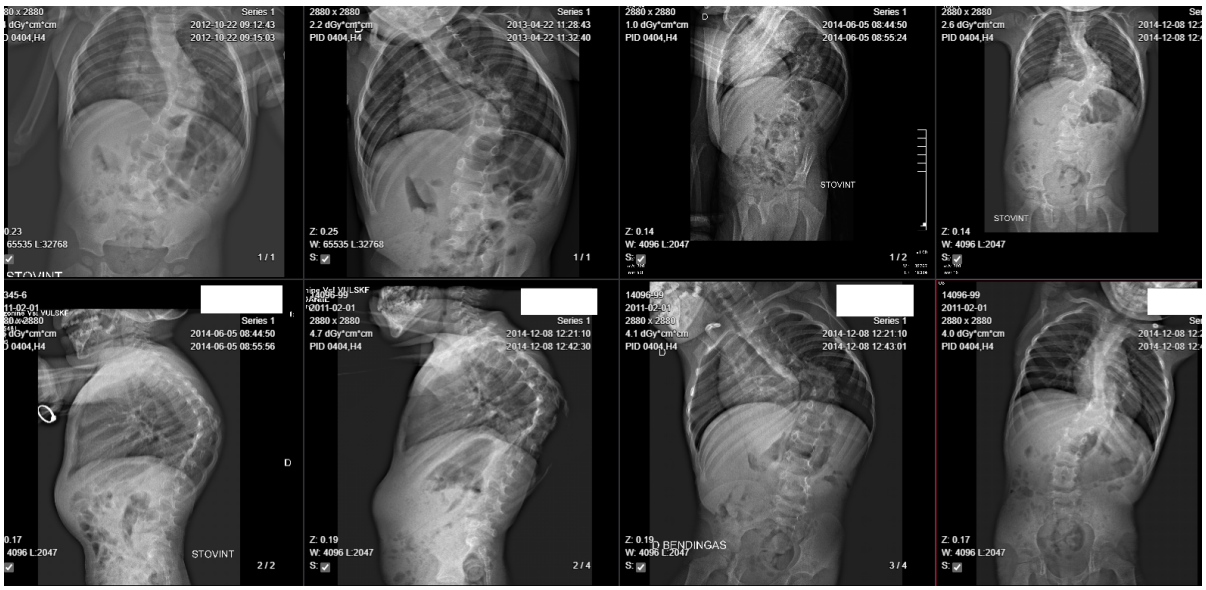

Figure 11: Serial lateral and AP radiographs during observation.

Without treatment, congenital scoliosis frequently leads to poor outcomes (20). Only a small minority of 10% of untreated patients will have a mild curve (≤20°), while the majority with 64% progress to severe deformities exceeding 40° (20). Figure 4 provides a guide to surgical decision-making in congenital scoliosis. In this guide, the treatment strategy depends on the number of malformed vertebral segments and whether additional thoracic abnormalities, like fused ribs, are present. In children with three or fewer malformed vertebral segments, a preventive approach or a corrective approach can be used. The preventive approach consists of in-situ fusion. This method is usually done in cases where the curvature is stable and not progressing rapidly. In situ fusion aims to halt further progression without attempting significant progression. Another method is the corrective approach which includes the hemivertebra resection or hemiepiphysiodesis. Hemivertebrectomy can immediately improve alignment and is labeled acute because correction occurs at the time of surgery. Alternatively, hemiepiphysiodesis is a gradual correction method.

When more than three segments are involved, the strategy shifts toward growth-friendly procedures like growth rods.

Some children with congenital scoliosis also have fused ribs, which can significantly impact thoracic growth and pulmonary function. If a patient presents with fused ribs, expansive thoracoplasty with a VEPTR device is indicated.

In our case, the surgical team selected a strategy called convex instrumented hemiepiphysiodesis with concave distraction. This decision was driven by the patient's young age, the presence of a long, multi-segmental thoracic curve, and the need to maintain spinal and thoracic growth.

Convex instrumented hemiepiphysiodesis with concave distraction applies growth-arresting pedicle screw instrumentation and simultaneously distracts the concave side over a growing rod construct

(35). An advantage of this technique is that it offers immediate improvement in coronal alignment and rotational control without the need for anterior fusion (35). Since it is a guided growth measure it also allows thoracic and spinal growth.

Compared to the other options, the convex instrumented hemiepiphysiodesis with concave distraction was deemed safer and more acceptable due to its anatomical complexity. Figures 12 and 13 display radiographic images before and after surgery.

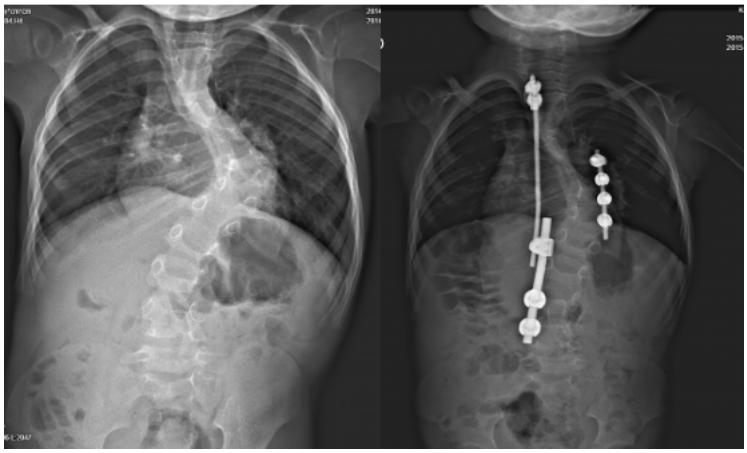

Figure 12: Comparison of AP radiographs before (left) and after (right) convex instrumented hemiepiphysiodesis with concave distraction.

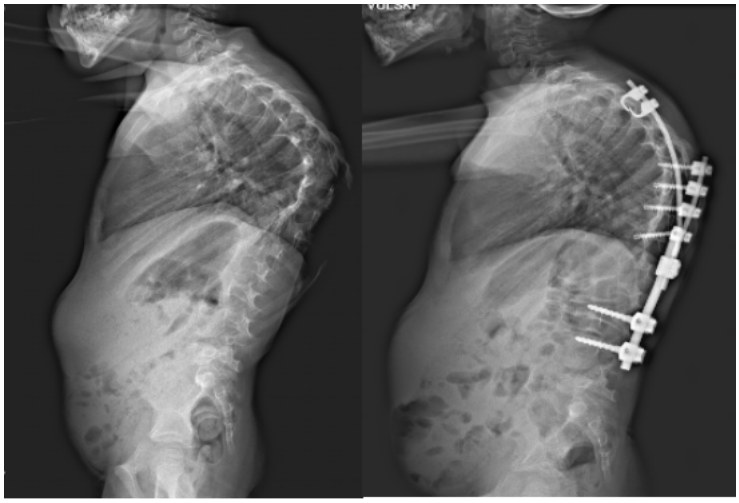

Figure 13: Comparison of lateral radiographs before (left) and after (right) convex instrumented hemiepiphysiodesis with concave distraction.

After the initial operation of a convex instrumented hemiepiphysiodesis with concave distraction, the patient entered a critical phase of follow-up, which lasted several years. Postoperative radiographs initially showed promising correction in both the coronal and sagittal planes, consistent with the expected outcome of this technique. (Image 14)

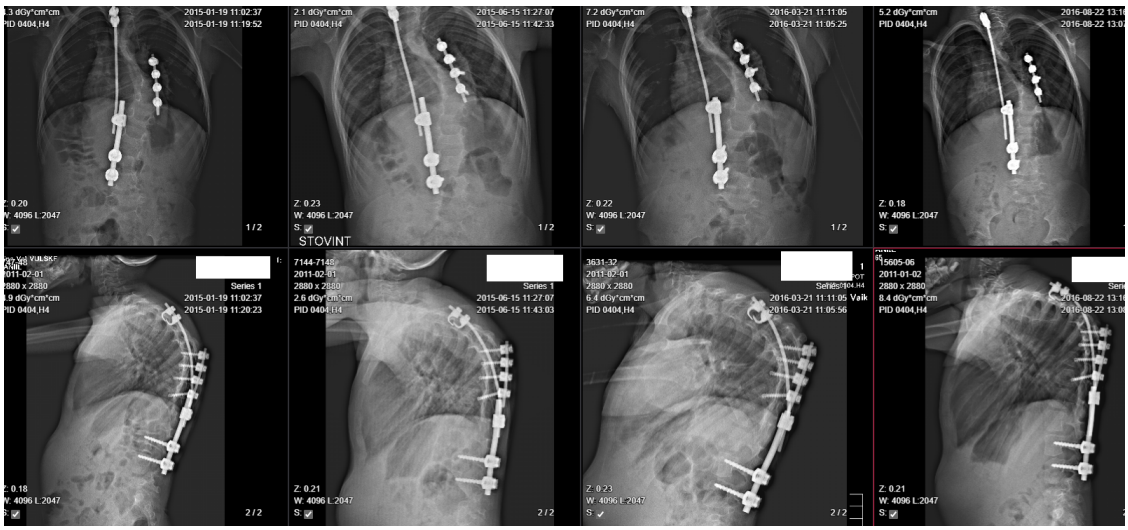

Figure 14: Serial AP and lateral radiographs during the postoperative follow-up.

Rod breakage, which occurred in our patient, is a well-documented complication of growth-friendly spinal instrumentation (Figure 15). It can significantly impact clinical outcomes and often necessitates unplanned revision surgeries. These fractures typically result from bending fatigue, especially under repeated flexion stress over time (36). Breakage tends to occur at biomechanically vulnerable locations, such as the apex of the deformity or the distraction junctions, where repetitive loading is mostly concentrated due to dynamic spinal motion and corrective distraction cycles (36). Additional insights into the clinical course of rod breakage are provided by Mehta et al., who evaluated mechanical failure in traditional growing rod constructs and found a mechanical failure rate of 26%, most often happening at the end of treatment (37). The study also noted that dual-rod constructs are more resilient than single-rod setups (37). In contrast, our case employed a convex instrumented hemiepiphysiodesis with concave distraction, a technique that avoids repeated lengthening. This approach offers a biomechanical advantage in long, stiff congenital curves by guiding asymmetric growth without the need for distraction-induced stress (35).

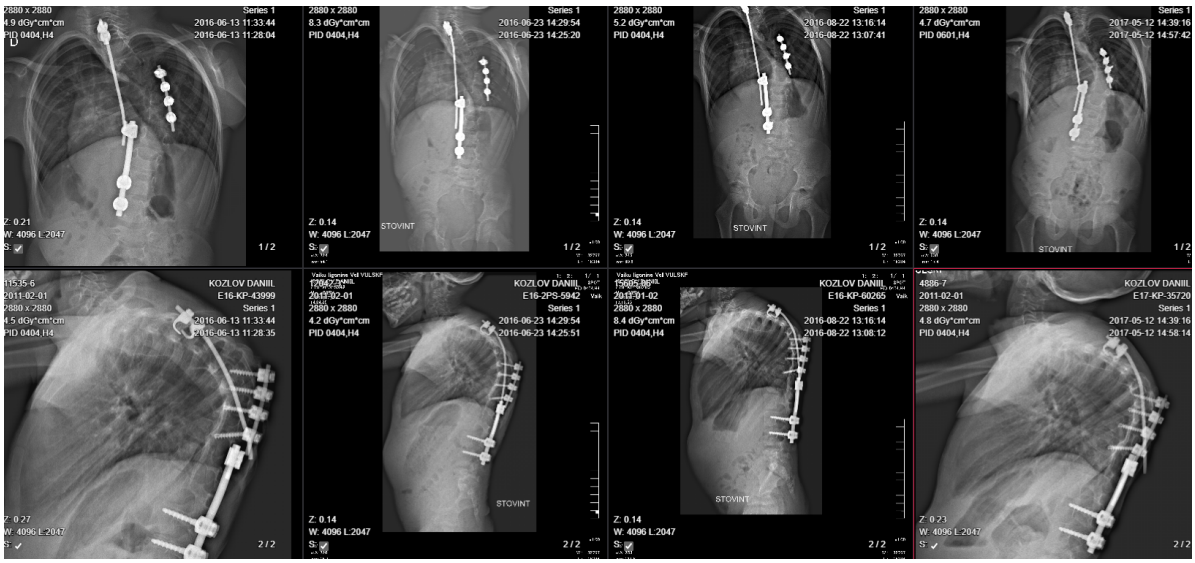

Figure 15: Serial AP and lateral radiographs showing rod breakage during follow-up.

During the patient's postoperative course, radiographs showed a dislocation of the upper implants (Figure 16). This complication is consistent with the known challenges of growth-friendly spinal instrumentation, particularly in constructs involving distraction and fusionless techniques.

Upper implant dislocation is a recognized complication in convex instrumented hemiepiphysiodesis with concave distraction, especially when single screws are used for fixation. Demirkiran et al. observed this issue in four out of 11 patients who were treated by this technique and in whom the proximal, distal, or both pedicle screws were partially pulled out (35). The use of single-level pedicle screws was identified as a risk factor, and the authors recommend extending fixation across two levels proximally and distally to reduce the risk of mechanical failure (35).

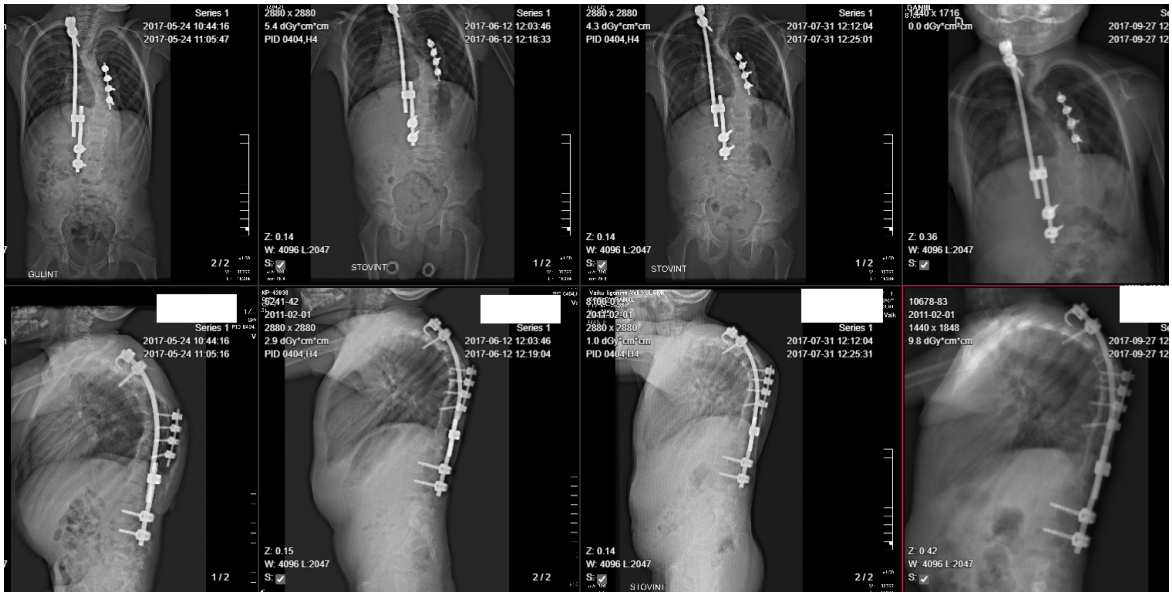

Figure 16: serial AP and lateral radiographs following revision surgery showing upper implant dislocation.

Following the previously documented implant dislocation, further imaging shows the patient after surgical revision (Figure 17). The placement of additional proximal pedicle screws can be seen, suggesting that the surgical team was proactive in addressing previous implant issues. This is consistent with the already mentioned recommendation described in the literature, which emphasizes the importance of extending fixation beyond a single level to reduce mechanical failure (35).
Furthermore, it shows that the hemiepiphysiodesis instrumentation has been revised in the following. Instead, a bilateral dual rod construct has been implemented, suggesting a transition in the treatment strategy. The implant revision with bilateral rods improves construct rigidity and reduces the risk of further rod fatigue. As mentioned dual rod systems are biomechanically more stable and associated with fewer mechanical complications compared to single rod constructs (37). There is no further evidence of screw loosening or rod fracture at this stage.

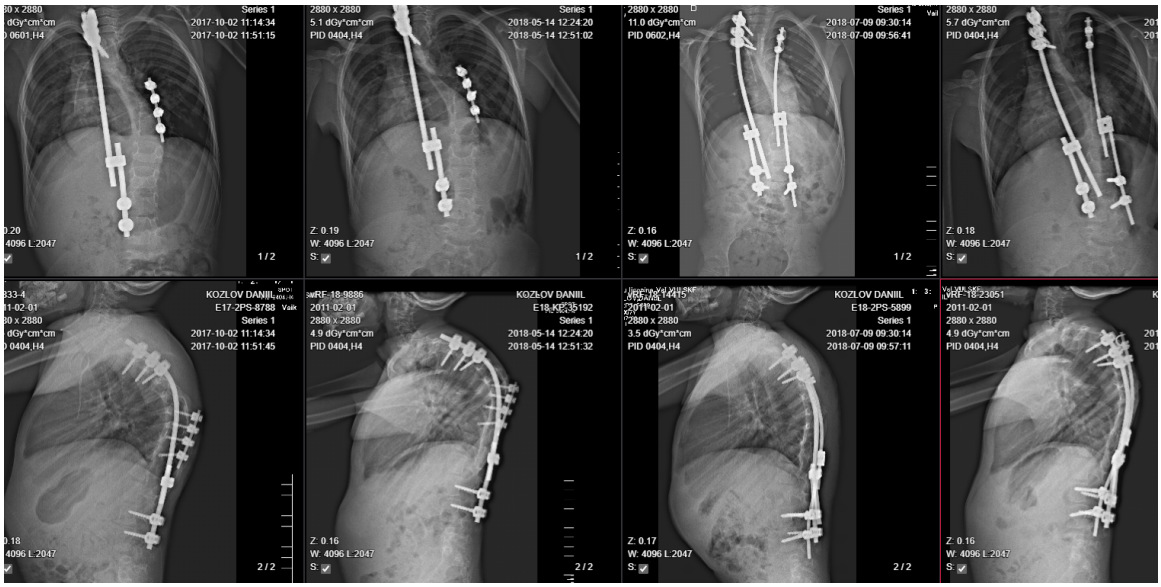

Figure 17: Final AP and lateral radiographs following definitive surgical correction with additional pedicle screws and the implementation of a bilateral rod construct.

In the following, a CT scan was done. It showed a hemivertebra at L5-S1, with apparent impingement of the spinal canal (Figure18). Despite this radiological finding, the patient remained neurologically intact at this stage. Nevertheless, such a finding is clinically significant, as vertebral malformations such as hemivertebrae can narrow the spinal canal and compromise the spinal cord or nerve roots over time. Although our patient did not exhibit neurological deterioration, the presence of impingement raises concern for potential tethering, nerve root irritation, or future compromise of motor and sensory function in the lower extremities. This scenario is reminiscent of the pathomechanics described in the case report by Hubbard et al., where a hemivertebra at T11 caused spinal cord compression, although in that case, the source was an arachnoid web (38). Although no arachnoid pathology was identified in our case, the mechanical narrowing from the L5-S1 vertebral

malformation may result in similar compressive effects, especially if left untreated or if the progression continues. Hubbard´s case underscores that even stable or long-standing deformities can later result in neural compromise, especially if structural anomalies narrow the canal (38). This reinforces the clinical decision in our case to proceed with surgical resection of the L5-S1 hemivertebra, even in the absence of immediate neurological deficits, as a preventive measure to avoid sudden or progressive neurological deterioration.

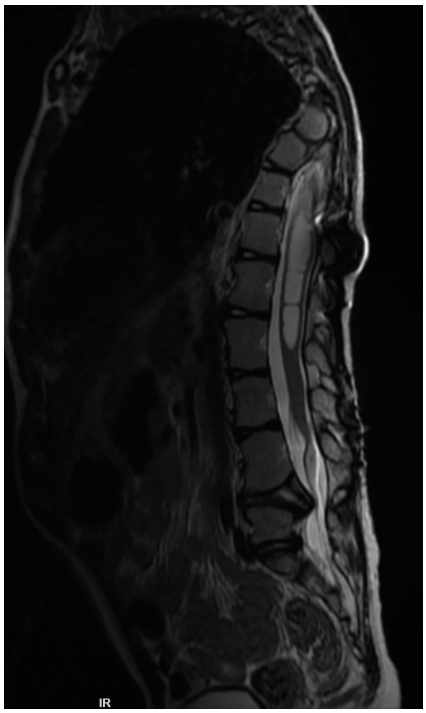

Figure 18: Sagittal T2-weighted MRI of the lumbar spine obtained during follow-up. The image shows impingement of the spinal canal at the L5-S1 level.

Following the identification of the hemivertebra at the L5-S1 level and the associated narrowing of the spinal canal, the decision was made to surgically resect the malformed vertebra. Although the patient had no neurological deficits at the time, the anatomical compression posed a clear risk of future complications. Given the lumbosacral location and the canal impingement, surgical resection was chosen to be the most effective preventive measure to protect long-term neurological and functional outcomes.

An important consideration in this case was the anatomical positioning of the malformation. The L5-S1 segment is positioned below the conus medullaris, meaning that the spinal cord has already transitioned into the cauda equina in this area; this makes it less risky for spinal cord injury (28). Before the definitive operation, the patient underwent a period of halo-gravity traction, lasting 3,5 weeks, with a force corresponding to approximately 50% of body weight. Halo traction is known to improve the flexibility of ridged curves and reduce spinal cord tension prior to major corrections. As

highlighted by Koller et al., this approach helps decrease neurological risk and optimize conditions for surgery by gradually stretching the spinal column (39). It also improves preoperative pulmonary function (39). In the context of this case, with an underlying impingement and prior syrinx formation, preoperative traction likely played a protective role in decompressing the spinal canal while allowing the surgical team to assess spinal cord tolerance in a controlled, low-risk manner.

Once traction achieved satisfactory correction, hemivertebra resection was performed. This approach involved the removal of the malformed vertebral body, along with its pedicles and adjacent discs. Gradual compression was applied using a posterior screw-rod system to close the osteotomy and stabilize the spine (40).

Following the hemivertebra resection, the patient developed radicular pain in the right leg consistent with L5 nerve root irritation. This likely resulted from mechanical stress or transient neural traction during posterior compression and instrumentation.

Given the complexity of the deformity and the child´s ongoing skeletal growth, the surgical team opted for a bilateral dual-rod construct spanning from Th1-S1. This arrangement, as stated before, provides improved stability, and better protection against mechanical fatigue compared to single-rod systems (37). The L5-S1 hemivertebra resection served both therapeutically and preventive. It decompressed structural impingement, addressed pelvic tilt, and corrected spinal alignment, all while minimizing neurologic risk. Most importantly, it was performed before the onset of irreversible deficits.

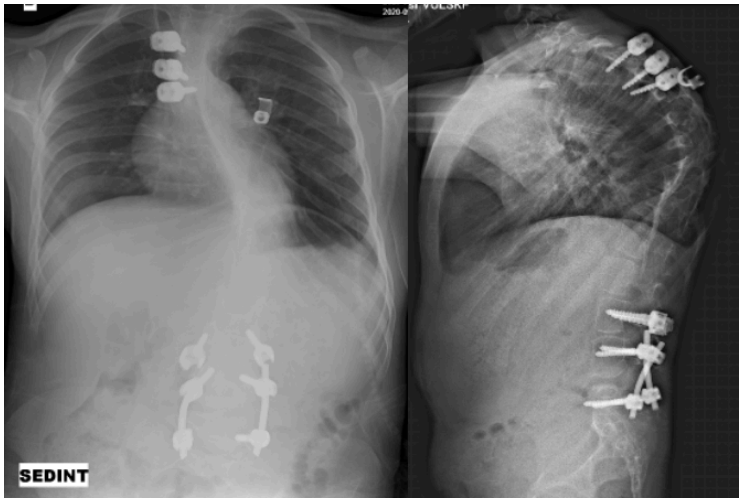

Figure 19: AP and lateral radiograph following revision surgery showing a newly implemented posterior srew-rod system.

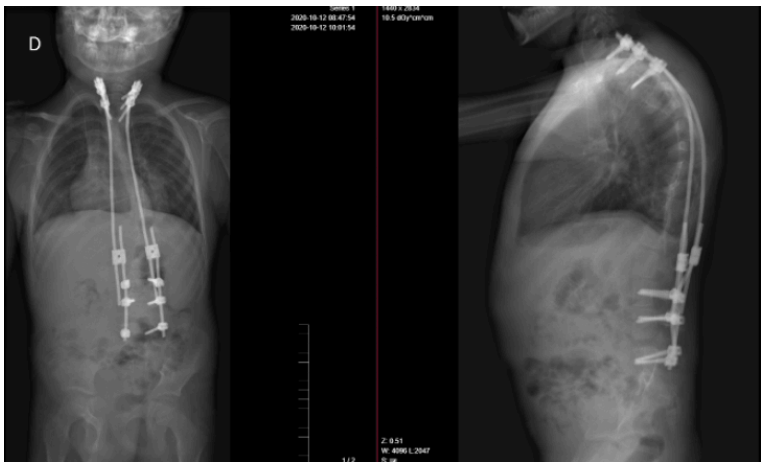

Figure 20: AP and lateral radiographs showing bilateral posterior screw-rod instrumentation.

Despite the successful resection of the L5-S1 hemivertebra, follow-up imaging of the sagittal profile revealed an insufficient restoration of global sagittal alignment. The patient continued to have a flat lumbar profile with insufficient lumbar lordosis. Considering the importance of the lumbar curvature in maintaining an upright posture and minimizing compensatory pelvic retroversion, correction of this sagittal imbalance was the next step in surgical management.

To achieve this, a pedicle subtraction osteotomy (PSO) was performed at the L3 level. This technique allows angular correction at a single vertebral level by removing a wedge-shaped segment of the vertebral body, including the pedicles and posterior elements, and is most commonly used in the lumbar region (41). PSO is particularly effective in rigid deformities, where intervertebral disc flexibility is limited, and was well-suited for this case due to the patient´s persistent hypolordosis and previous long-segment instrumentation (41). The L3 vertebra was chosen as the osteotomy site due to its central position in the lumbar spine and its frequent use in sagittal plane correction. According to Eskilsson et al., L3 and L2 are the most frequently selected levels for PSO, offering effective angular correction without requiring anterior release (42).

The rationale behind performing PSO was not only structural but also functional. If uncorrected, persistent sagittal imbalance is known to cause compensatory mechanisms such as pelvic retroversion, pain, increased energy expenditure during walking, and ultimately, functional disability and poor quality of life (41). It has been shown that when the sagittal alignment of the spine is restored within physiological parameters, long-term outcomes are significantly improved (41).

In this case, not only was the spinal profile permanently corrected and the spinopelvic harmony achieved with the combination of the L3 PSO and iliac screw fixation, but the long-term mechanical stability of the implant is further enhanced. The postoperative radiographs demonstrated restitution of the lumbar curvature and a well-balanced sagittal profile, indicative of the success of this global surgical strategy.

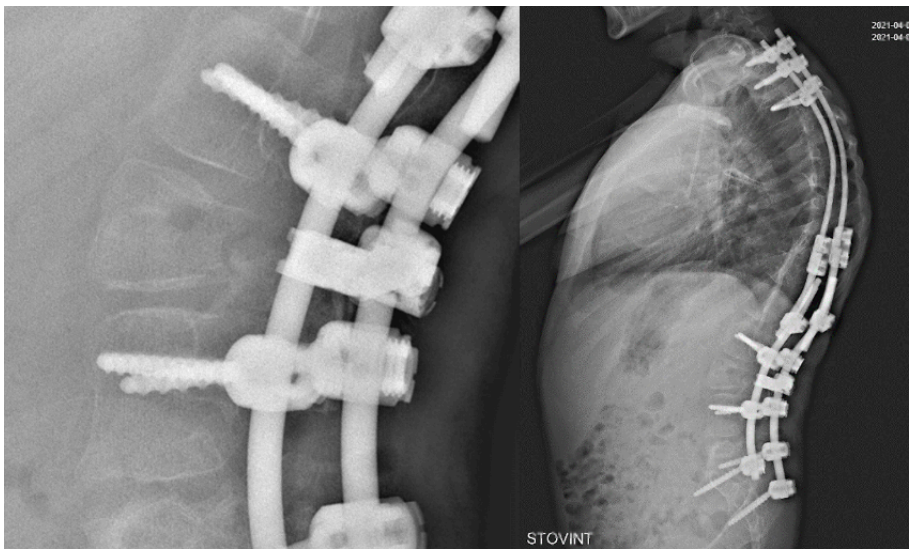

Figure 21: Detailed lateral view of the lumbosacral instrumentation (left) and overall sagittal alignment after final correction (right).

7. Discussion

The literature provides numerous recommendations and decision-making algorithms in the treatment of CS, but this case also shows that standard protocols often reach their limits and require customization. Its treatment is a major challenge for the surgeon due to the complexity and individuality of each case. A very thorough examination of the patient is necessary, as well as close follow-up monitoring, as the complication rate is very high at about 50%.

The findings presented in this work are in line with scientific understanding, which is becoming increasingly interdisciplinary and differentiated.

The 13-month-old boy with thoracic kyphoscoliosis, syrinx, and an additional lumbar hemivertebra which was described in this case, is an example of how multidisciplinary thinking, precise pre-, intra-, and postoperative imaging and an individualized surgical concept must be combined.

Armer Seebaly and colleagues proposed a very detailed and structured approach for the appropriate treatment of congenital scoliosis, which is shown in Figure 5.

The decision tree is divided into two major categories which are children younger than six years and children ages six years until puberty. In infants and young children under six years, early detection and monitoring are essential since congenital scoliosis may progress rapidly. The treatment depends primarily on the Cobb angle and whether the child shows signs of TIS. For cases where the CA does not exceed 40°, the recommended approach is regular radiographic follow-up to monitor the curve progression. X-rays are performed every three months, and if the curve progresses by less than 5°, the interval increases to all six and all twelve months. If the curve progresses by more than 5°, X-rays are performed every three months. A surgical intervention is required if the CA is equal to or exceeds 40°. If the patient is older than six months and there is a type II rib deformity, VEPTR is indicated. It

is also indicated if the patient is older than three years and thoracic insufficiency syndrome is present. If the CA is between 40° and 50° and the patient has no lordosis in the region, a posterior in situ fusion is indicated. With a CA ≥ 50° and a hemivertebra at the apex, a hemivertebrectomy is indicated. In the case of a fully segmented vertebra and a CA under 70° and a sagittal angle between 10° and 50°, a hemiepiphysiodesis with or without a posterior concavity distraction or Growing Rods are indicated. Since our patient is 13 months old, we will not discuss the approach for patients older than six.

Following this treatment algorithm, two different surgery options are available and could be used.

The first one is the hemivertebra resection since our patient has a CA over 50° and a hemivertebra at the apex. The second one is the hemiepiphysiodesis with or without posterior concavity distraction or growing rods because the patient has a fully segmented hemivertebra and a CA under 70° but the patient has a kyphosis over 50° which speaks against it.

Compared to the other options, the convex instrumented hemiepiphysiodesis with concave distraction was deemed safer and more acceptable due to its anatomical complexity.

Hemivertebra resection was ruled out due to several clinical factors. First, the patient presented with a multilevel thoracic deformity, including a hemivertebra at T8, thoracic kyphosis exceeding 70°, and the presence of a spinal cord syrinx extending from T2 to L1. Although hemivertebra resection is an established procedure for the correction of isolated congenital deformities, it is mainly indicated for patients with single and two-level vertebral defects, particularly in the thoracolumbar and lumbar spine (2,20). In addition, hemivertebral excision often requires adjacent-level fusion to stabilize the spine, which may compromise future thoracic and spinal growth. This limitation is even more pronounced in the thoracic spine, where extensive fusion may compromise lung development (35).

In addition, resection in the presence of a syrinx increases the risk of neurological injury. The literature on scoliosis with syringomyelia emphasizes the need for caution in instrumentation and resection in such cases, as the spinal cord is vulnerable (34). The presence of a large syrinx can, therefore, be a factor in excluding hemivertebra resection, as this requires precise manipulation near the cord.

Growing rods can also be considered but were excluded since the ridged, malformed curve makes it unlikely that growing rods alone would control the deformity at the apex (35).

In-situ fusion was ruled out due to its known effect on halting spinal and thoracic development if used in a multi-level segment (2). The patient's curve measured 53°, exceeding the limit typically considered acceptable in in-situ fusion (2).

Finally, VEPTR can be excluded in this case as the patient does not have fused ribs or TIS.

In conclusion, convex instrumented hemiepiphysiodesis with concave distraction offered the best compromise between correction and growth preservation. It is particularly effective in treating long,

ridged curves in very young children while avoiding the risks associated with more aggressive resection or fusion techniques (35).

Another important point in the diagnosis and treatment of congenital scoliosis that was highlighted in this case is the importance of imaging. Firstly, preoperatively, a syrinx was detected, with 5-20,2% quite common in patients with congenital scoliosis, illustrating the importance of preoperative imaging (2). Its discovery was important for planning the surgical strategy and indicated the need for a shunt.

Follow-up imaging revealed complications, in our case, a rod breakage and an upper implant dislocation, which happens in approximately 50% of all patients with growing rods (5). Due to this discovery, a later revision was necessary.

During the course of the disease, magnetic resonance imaging revealed a new stenosis at the L5-S1 level, although the patient initially had no neurological abnormalities. Hemivertebrectomy was subsequently required. In addition, further sagittal radiographs revealed inadequate lumbar lordosis, necessitating pedicle subtraction osteotomy (PSO). These developments highlight the importance of continuous multimodal imaging as a key tool in the long-term management of complex congenital scoliosis.

A promising approach to optimising these imaging-based processes is the integration of artificial intelligence (AI). Modern AI-based systems are already able to automatically and accurately analyse parameters such as the Cobb angle, which can be particularly helpful in the context of follow-up care (43). In addition, machine learning algorithms allow for early detection of complex deformities based on image data and can thus contribute to risk stratification and treatment planning (43,44). Although these technologies have mainly been evaluated in idiopathic scoliosis, their future use in congenital scoliosis seems particularly useful due to the complexity of the imaging.

The use of patient-specific 3D-printed spinal models is another technological enhancement that can be particularly beneficial in anatomically challenging conditions, as in the case described here involving multiple malformations.

These offer the surgical team a real, haptic reference structure that permits even more precise planning and intraoperative guidance (45). A case study showed that such 3D models enable better visualization of the pathoanatomical conditions than conventional CT or MRI data alone (45).

This technique is particularly suitable for complex pathologies and morphologies (45).

An individualized 3D model could also have potentially helped to minimize the risk in our patient, for example, through more precise planning of screw positioning or segment selection during stabilization surgery.

In addition to traditional imaging, innovative technologies such as AI-based image analysis and 3D-printed anatomical models are becoming increasingly important. Their targeted use in complex cases such as the one described here could lead to more precise decisions, better functional results, and possibly lower revision rates in the future.

Besides further technological development, a deeper understanding of molecular biological principles is also important to better predict and treat complex courses such as the case presented here. Even though no genetic or epigenetic diagnostics were carried out on our patient, current research results indicate that epigenetic regulatory processes, such as DNA methylation, histone modifications, and non-coding RNAs, play a significant role in the development of forms of scoliosis, including congenital scoliosis (46).
Given the pronounced deformity with multiple hemivertebrae and an initial Cobb angle of over 50°, it is a possibility that epigenetic dysregulation also contributed to the pathogenesis in this case. A study by Wu et al. showed that the degree of methylation level of the KAT6B gene was positively correlated with the Cobb angle (47). However, according to the current literature, there is still a considerable need for research, particularly in the area of chromatin remodeling and histone modification, as these mechanisms have not been insufficiently investigated in the context of congenital scoliosis (46).
At the same time, the authors point out that epigenetic changes are, in principle, reversible, which could open up new approaches to future diagnostic or therapeutic strategies (46).

Alongside the surgical complexity and careful imaging-based planning, the quality of life of the affected children and their families is an equally central aspect in the treatment of congenital scoliosis. Functional limitations and visible deformity can have a negative impact on both the child´s physical performance and self-image. A study by Hsu et al. shows that in particular the subjective sense of health is impaired in patients with congenital scoliosis, while other areas such as physical function, social interaction and emotional well-being remain comparatively stable (48). The family environment is also under considerable stress, particularly in the early post-diagnosis period. A systematic review by Motyer et al. shows that parents are often confronted with a flood of information, unclear prognoses and significant emotional stress, which can have an immediate negative impact on their mental health (49).
It is likely that the multi-step nature of the patient's illness, with repeated interventions, regular imaging and revision surgery, has also placed a heavy emotional burden on the family. At the same time, it can be assumed that the structured surgical planning and the visible improvements in the postoperative course contributed to psychological relief. Modern technological aids such as the

patient's specific 3D model can also indirectly have a positive impact on quality of life, for example, through improved preoperative visualization, which supports the medical team in decision-making and also contributes to a better understanding among parents (45).

8. Conclusion

Congenital scoliosis is a complex and clinically significant malformation of the spinal column that arises from complex malformation mechanisms in early embryogenesis and is often accompanied by systemic malformations. The diagnostic and therapeutic care of affected children requires a high degree of interdisciplinary cooperation as well as an individual and dynamic treatment strategy, as the course and prognosis are strongly influenced by the type, number, and localization of the vertebral body anomalies as well as the possible comorbidities. A central aim of modern pediatric spinal surgery is not only to correct the deformity but also to prevent long-term complications, such as thoracic insufficiency syndrome.

The combination of different surgical approaches, ranging from shunt implantation to treat the syrinx to hemivertebral resection and pedicle substraction osteotomy to restore sagittal balance, shows that the success of treatment is largely dependent on continuous re-evaluation and adaptation of surgical planning. Repeated imaging was crucial not only for the identification of acute complications but also for the early detection of clinically relevant secondary pathologies.

This case also illustrates the enormous psychological and logistical burden of long-term therapy for both the affected child and the family. It emphasizes the need to incorporate psychosocial aspects into care at an early stage alongside medical and surgical standards. The potential contribution of new technologies, such as AI-based image analysis or patient-specific three-dimensional print models, to optimize planning and information and reduce complications offers forward-looking opportunities here.

Even though genetic or epigenetic diagnostics were not performed in the case presented, current research findings emphasize the relevance of epigenetic regulatory mechanisms in the pathogenesis of congenital scoliosis. Their potential reversibility opens up long-term prospects for new diagnostic and therapeutic approaches. In addition to technical precision, this also requires foresighted action, individual consideration, and the willingness to integrate new technologies and findings into the therapeutic concept in a targeted manner.

9. Sources

1. David Abbasi JS. Congenital Scoliosis - Spine - Orthobullets [Internet]. [cited 2025 Mar 14]. Available from: https://www.orthobullets.com/spine/2060/congenital-scoliosis
2. Sebaaly A, Daher M, Salameh B, Ghoul A, George S, Roukoz S. Congenital scoliosis: a narrative review and proposal of a treatment algorithm. EFORT Open Rev. 2022 May 5;7(5):318-327. doi: 10.1530/EOR-21-0121. PMID: 35510738; PMCID: PMC9142820.
3. Furdock R, Brouillet K, Luhmann SJ. Organ System Anomalies Associated With Congenital Scoliosis: A Retrospective Study of 305 Patients. J Pediatr Orthop. 2019 Mar;39(3):e190-e194. doi: 10.1097/BPO.0000000000001279. PMID: 30376499.
4. Fekete TF, Haschtmann D, Heyde CE, Kleinstück F, Jeszenszky D. Kongenitale Fehlbildungen der Wirbelsäule im Wachstum : Wann konservativ behandeln, wann operativ? [Congenital malformations of the growing spine : When should treatment be conservative and when should it be surgical?]. Orthopade. 2016 Jun;45(6):518-26. German. doi: 10.1007/s00132-016-3275-4. PMID: 27225659.
5. Mackel CE, Jada A, Samdani AF, Stephen JH, Bennett JT, Baaj AA, Hwang SW. A comprehensive review of the diagnosis and management of congenital scoliosis. Childs Nerv Syst. 2018 Nov;34(11):2155-2171. doi: 10.1007/s00381-018-3915-6. Epub 2018 Aug 4. PMID: 30078055.
6. Cho W, Shepard N, Arlet V. The etiology of congenital scoliosis: genetic vs. environmental-a report of three monozygotic twin cases. Eur Spine J. 2018 Jul;27(Suppl 3):533-537. doi: 10.1007/s00586-018-5604-2. Epub 2018 Apr 18. PMID: 29671107.
7. Magli A, Schnettler E, Rinaldi F, Bremer P, Perlingeiro RC. Functional dissection of Pax3 in paraxial mesoderm development and myogenesis. Stem Cells. 2013 Jan;31(1):59-70. doi: 10.1002/stem.1254. PMID: 23081715; PMCID: PMC3528848.
8. Wu N, Ming X, Xiao J, Wu Z, Chen X, Shinawi M, Shen Y, Yu G, Liu J, Xie H, Gucev ZS, Liu S, Yang N, Al-Kateb H, Chen J, Zhang J, Hauser N, Zhang T, Tasic V, Liu P, Su X, Pan X, Liu C, Wang L, Shen J, Shen J, Chen Y, Zhang T, Zhang J, Choy KW, Wang J, Wang Q, Li S, Zhou W, Guo J, Wang Y, Zhang C, Zhao H, An Y, Zhao Y, Wang J, Liu Z, Zuo Y, Tian Y, Weng X, Sutton VR, Wang H, Ming Y, Kulkarni S, Zhong TP, Giampietro PF, Dunwoodie SL, Cheung SW, Zhang X, Jin L, Lupski JR, Qiu G, Zhang F. TBX6 null variants and a common hypomorphic allele in congenital scoliosis. N Engl J Med. 2015 Jan 22;372(4):341-50. doi: 10.1056/NEJMoa1406829. Epub 2015 Jan 7. PMID: 25564734; PMCID: PMC4326244.
9. Chen W, Liu J, Yuan D, Zuo Y, Liu Z, Liu S, Zhu Q, Qiu G, Huang S, Giampietro PF, Zhang F, Wu N, Wu Z. Progress and perspective of TBX6 gene in congenital vertebral malformations.

Oncotarget. 2016 Aug 30;7(35):57430-57441. doi: 10.18632/oncotarget.10619. PMID: 27437870; PMCID: PMC5302999.

10. Takeda K, Kou I, Mizumoto S, Yamada S, Kawakami N, Nakajima M, Otomo N, Ogura Y, Miyake N, Matsumoto N, Kotani T, Sudo H, Yonezawa I, Uno K, Taneichi H, Watanabe K, Shigematsu H, Sugawara R, Taniguchi Y, Minami S, Nakamura M, Matsumoto M; Japan Early Onset Scoliosis Research Group; Watanabe K, Ikegawa S. Screening of known disease genes in congenital scoliosis. Mol Genet Genomic Med. 2018 Nov;6(6):966-974. doi: 10.1002/mgg3.466. Epub 2018 Sep 9. PMID: 30196550; PMCID: PMC6305645.
11. Burnei G, Gavriliu S, Vlad C, Georgescu I, Ghita RA, Dughilă C, Japie EM, Onilă A. Congenital scoliosis: an up-to-date. J Med Life. 2015 Jul-Sep;8(3):388-97. PMID: 26351546; PMCID: PMC4556925.
12. Deml MC, Tinner C, Albers CE. Klinische und radiologische Untersuchung der idiopathischen Skoliose. Paediatrica [Internet]. 2023 Dec 20 [cited 2025 Apr 8];34(4). Available from: https://www.paediatrieschweiz.ch/klinische-und-radiologische-untersuchung-skoliose/
13. Hogue GD, Emans JB. Thoracic Insufficiency Syndrome. J Pediatr Orthop Soc N Am. 2021 Aug;3(3):311.
14. Horng MH, Kuok CP, Fu MJ, Lin CJ, Sun YN. Cobb Angle Measurement of Spine from X-Ray Images Using Convolutional Neural Network. Comput Math Methods Med. 2019 Feb 19;2019:6357171. doi: 10.1155/2019/6357171. PMID: 30996731; PMCID: PMC6399566.
15. Misbah I, Ravula P, Raja S, Mohanakrishnan A, Natarajan P, Gunasekaran D. Comprehensive Radiological Imaging for the Characterization of Spinal Dysraphism and Associated Anomalies in a Pediatric Case. Cureus. 2024 Sep 1;16(9):e68415. doi: 10.7759/cureus.68415. PMID: 39360046; PMCID: PMC11446498.
16. Przybylski R, Hedequist DJ, Nasr VG, McCann ME, Brustowicz RM, Emans JB, Marshall AC, Brown DW. Adverse Perioperative Events in Children with Complex Congenital Heart Disease Undergoing Operative Scoliosis Repair in the Contemporary Era. Pediatr Cardiol. 2019 Oct;40(7):1468-1475. doi: 10.1007/s00246-019-02169-1. Epub 2019 Jul 26. PMID: 31350568.
17. Ghiță RA, Georgescu I, Muntean ML, Hamei Ș, Japie EM, Dughilă C, Țiripa I. Burnei-Gavriliu classification of congenital scoliosis. J Med Life. 2015 Apr-Jun;8(2):239-44. PMID: 25866585; PMCID: PMC4392099.
18. Pahys JM, Guille JT. What's New in Congenital Scoliosis? J Pediatr Orthop. 2018 Mar;38(3):e172-e179. doi: 10.1097/BPO.0000000000000922. PMID: 28009797.
19. Negrini S, Donzelli S, Aulisa AG, Czaprowski D, Schreiber S, de Mauroy JC, Diers H, Grivas TB, Knott P, Kotwicki T, Lebel A, Marti C, Maruyama T, O'Brien J, Price N, Parent E, Rigo M, Romano M, Stikeleather L, Wynne J, Zaina F. 2016 SOSORT guidelines: orthopaedic and

rehabilitation treatment of idiopathic scoliosis during growth. Scoliosis Spinal Disord. 2018 Jan 10;13:3. doi: 10.1186/s13013-017-0145-8. PMID: 29435499; PMCID: PMC5795289.

20. Tikoo A, Kothari MK, Shah K, Nene A. Current Concepts - Congenital Scoliosis. Open Orthop J. 2017 Apr 28;11:337-345. doi: 10.2174/1874325001711010337. PMID: 28603565; PMCID: PMC5447938.
21. Ashebo L, Anari JB, Cahill PJ. Update on the Diagnosis and Management of Early-onset Scoliosis. Curr Rev Musculoskelet Med. 2023 Oct;16(10):447-456. doi: 10.1007/s12178-023-09848-w. Epub 2023 Aug 24. PMID: 37615932; PMCID: PMC10497459.
22. Latalski M, Fatyga M, Sowa I, Wojciak M, Starobrat G, Danielewicz A. Complications in growth-friendly spinal surgeries for early-onset scoliosis: Literature review. World J Orthop. 2021 Aug 18;12(8):584-603. doi: 10.5312/wjo.v12.i8.584. PMID: 34485105; PMCID: PMC8384615.
23. Gavriliu S, Sora EM. The Use of VEPTR in Congenital Scoliosis.
24. Asunis E, Cini C, Martikos K, Vommaro F, Evangelisti G, Griffoni C, Gasbarrini A. Efficacy and Risks of Posterior Vertebral Column Resection in the Treatment of Severe Pediatric Spinal Deformities: A Case Series. J Clin Med. 2025 Jan 9;14(2):374. doi: 10.3390/jcm14020374. PMID: 39860381; PMCID: PMC11766027.
25. Hesemann J, Lauer E, Ziska S, Noonan K, Nemeth B, Scott-Schwoerer J, McCarty C, Rasmussen K, Goldberg JM, Sund S, Eickhoff J, Raggio CL, Giampietro PF. Analysis of maternal risk factors associated with congenital vertebral malformations. Spine (Phila Pa 1976). 2013 Mar 1;38(5):E293-8. doi: 10.1097/BRS.0b013e318283be6e. PMID: 23446706; PMCID: PMC3959640.
26. Chen JW, Koester SW, Liles C, Gannon S, Bonfield CM. Evaluating the prevalence of psychiatric comorbidities associated with pediatric scoliosis utilizing ResearchMatch. Spine Deform. 2024 Nov;12(6):1583-1593. doi: 10.1007/s43390-024-00926-8. Epub 2024 Aug 12. PMID: 39134889; PMCID: PMC11499428.
27. Tauchi R, Tsuji T, Cahill PJ, Flynn JM, Flynn JM, Glotzbecker M, El-Hawary R, Heflin JA, Imagama S, Joshi AP, Nohara A, Ramirez N, Roye DP Jr, Saito T, Sawyer JR, Smith JT, Kawakami N. Reliability analysis of Cobb angle measurements of congenital scoliosis using X-ray and 3D-CT images. Eur J Orthop Surg Traumatol. 2016 Jan;26(1):53-7. doi: 10.1007/s00590-015-1701-7. Epub 2015 Sep 16. PMID: 26377663.
28. Department of Orthopaedics, Mashhad University of Medical Sciences, Emdadi Kamyab Hospital, Mashhad, Iran., Bagheri F, Razi A, Department of Orthopaedics, Mashhad University of Medical Sciences, Emdadi Kamyab Hospital, Mashhad, Iran., Birjandinejad A, Department of Orthopaedics, Mashhad University of Medical Sciences, Emdadi Kamyab Hospital, Mashhad,

Iran., et al. Congenital Scoliosis: A Current Concepts Review. J Pediatr Rev. 2021 Apr 1;9(2):127–36.

29. Mehta MH. The rib-vertebra angle in the early diagnosis between resolving and progressive infantile scoliosis. J Bone Joint Surg Br. 1972 May;54(2):230-43. PMID: 5034823.
30. Omara C, Pieters L, Castelein RM, Sakkers RJB, Vleggeert-Lankamp CLA. Early evaluation and treatment of thoracolumbar kyphosis in children with achondroplasia. Eur Spine J. 2025 Feb 3. doi: 10.1007/s00586-025-08692-5. Epub ahead of print. PMID: 39894831.
31. Xu HF, Li C, Ma ZS, Wu ZX, Sha J, Diwu WL, Yan YB, Liu ZC, Fan ZZ, Huang LY. Closing-opening wedge osteotomy for the treatment of congenital kyphosis in children. J Orthop Surg (Hong Kong). 2022 Sep-Dec;30(3):10225536221118600. doi: 10.1177/10225536221118600. PMID: 36120861.
32. Mohanty SP, Kanhangad MP, Saifuddin S, Narayana Kurup JK. Pattern of Syringomyelia in Presumed Idiopathic and Congenital Scoliosis. Asian Spine J. 2021 Dec;15(6):791-798. doi: 10.31616/asj.2020.0216. Epub 2020 Nov 16. PMID: 33189109; PMCID: PMC8696068.
33. Shenoy VS, Munakomi S, Sampath R. Syringomyelia. 2024 Mar 14. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan–. PMID: 30725795.
34. Zhu C, Huang S, Song Y, Liu H, Liu L, Yang X, Zhou C, Hu B, Chen H. Surgical Treatment of Scoliosis-Associated with Syringomyelia: The Role of Syrinx Size. Neurol India. 2020 Mar-Apr;68(2):299-304. doi: 10.4103/0028-3886.280648. PMID: 32189709.
35. Demirkiran G, Dede O, Ayvaz M, Bas CE, Alanay A, Yazici M. Convex Instrumented Hemiepiphysiodesis With Concave Distraction: A Treatment Option for Long Sweeping Congenital Curves. J Pediatr Orthop. 2016 Apr-May;36(3):226-31. doi: 10.1097/BPO.0000000000000441. PMID: 25812144.
36. Hill G, Nagaraja S, Akbarnia BA, Pawelek J, Sponseller P, Sturm P, Emans J; Growing Spine Study Group; Bonangelino P, Cockrum J, Kane W, Dreher M. Retrieval and clinical analysis of distraction-based dual growing rod constructs for early-onset scoliosis. Spine J. 2017 Oct;17(10):1506-1518. doi: 10.1016/j.spinee.2017.04.020. Epub 2017 Apr 26. PMID: 28456673.
37. Mehta JS, Tognini M, Hothi H. Growing rods in Early Onset Scoliosis: The current scenario. J Orthop. 2023 Jul 17;42:63-69. doi: 10.1016/j.jor.2023.07.008. PMID: 37519912; PMCID: PMC10372050.
38. Hubbard ME, Hunt MA, Jones KE, Polly DW. Thoracic spinal cord impingement by an arachnoid web at the level of a hemivertebra: case report. J Neurosurg Spine. 2017 Dec;27(6):638-642. doi: 10.3171/2017.5.SPINE161243. Epub 2017 Sep 8. PMID: 28885124.
39. Koller H, Zenner J, Gajic V, Meier O, Ferraris L, Hitzl W. The impact of halo-gravity traction on curve rigidity and pulmonary function in the treatment of severe and rigid scoliosis and

kyphoscoliosis: a clinical study and narrative review of the literature. Eur Spine J. 2012 Mar;21(3):514-29. doi: 10.1007/s00586-011-2046-5. Epub 2011 Nov 1. PMID: 22042044; PMCID: PMC3296862.

40. Li H, Chen Z, Gao B, Wang J, Shao S, Wu J. Surgical outcomes in children under 10 years old in the treatment of congenital scoliosis due to single nonincarcerated thoracolumbar hemivertebra: according to the age at surgery. J Orthop Surg Res. 2021 Dec 20;16(1):721. doi: 10.1186/s13018-021-02865-w. PMID: 34930346; PMCID: PMC8686336.
41. Falkowski GA, Labaran L, Hassanzadeh H. Sagittal alignment and thoracolumbar osteotomies. Semin Spine Surg. 2019 Jun;31(2):61–9.
42. Eskilsson K, Sharma D, Johansson C, Hedlund R. Pedicle subtraction osteotomy: a comprehensive analysis in 104 patients. Does the cause of deformity influence the outcome? J Neurosurg Spine. 2017 Jul;27(1):56-62. doi: 10.3171/2016.12.SPINE16585. Epub 2017 May 5. PMID: 28475018.
43. Zhang H, Huang C, Wang D, Li K, Han X, Chen X, Li Z. Artificial Intelligence in Scoliosis: Current Applications and Future Directions. J Clin Med. 2023 Nov 29;12(23):7382. doi: 10.3390/jcm12237382. PMID: 38068444; PMCID: PMC10707441.
44. Ohyama S, Maki S, Kotani T, Ogata Y, Sakuma T, Iijima Y, Akazawa T, Inage K, Shiga Y, Inoue M, Arai T, Toshi N, Tokeshi S, Okuyama K, Tashiro S, Suzuki N, Eguchi Y, Orita S, Minami S, Ohtori S. Machine learning algorithms for predicting future curve using first and second visit data in female adolescent idiopathic scoliosis patients. Eur Spine J. 2025 Feb 4. doi: 10.1007/s00586-025-08680-9. Epub ahead of print. PMID: 39903251.
45. Perfetti DC, Kisinde S, Belanger TA, Lieberman IH. Perioperative Benefits of a 3D Printed Spine Biomodel in the Setting of Congenital Scoliosis Surgery. Surg Tech Dev. 2024 Aug 9;13(3):278–93.
46. Sun D, Ding Z, Hai Y, Cheng Y. Advances in epigenetic research of adolescent idiopathic scoliosis and congenital scoliosis. Front Genet. 2023 Jul 26;14:1211376. doi: 10.3389/fgene.2023.1211376. PMID: 37564871; PMCID: PMC10411889.
47. Wu Y, Zhang H, Tang M, Guo C, Deng A, Li J, Wang Y, Xiao L, Yang G. High methylation of lysine acetyltransferase 6B is associated with the Cobb angle in patients with congenital scoliosis. J Transl Med. 2020 May 24;18(1):210. doi: 10.1186/s12967-020-02367-z. PMID: 32448279; PMCID: PMC7245753.
48. Hsu PC, Feng CK, Huang SH, Chiu JW, Chou CL, Yang TF. Health-related quality of life in children and adolescent with different types of scoliosis: A cross-sectional study. J Chin Med Assoc. 2019 Feb;82(2):161-165. doi: 10.1097/JCMA.0000000000000020. PMID: 30839509.

49. Motyer G, Dooley B, Kiely P, Fitzgerald A. Parents' information needs, treatment concerns, and psychological well-being when their child is diagnosed with adolescent idiopathic scoliosis: A systematic review. Patient Educ Couns. 2021 Jun;104(6):1347-1355. doi: 10.1016/j.pec.2020.11.023. Epub 2020 Nov 25. PMID: 33280964.
50. Kawakami N, Tsuji T, Imagama S, Lenke LG, Puno RM, Kuklo TR; Spinal Deformity Study Group. Classification of congenital scoliosis and kyphosis: a new approach to the three-dimensional classification for progressive vertebral anomalies requiring operative treatment. Spine (Phila Pa 1976). 2009 Aug 1;34(17):1756-65. doi: 10.1097/BRS.0b013e3181ac0045. PMID: 19644327.