Congenital Scoliosis

Konjenital Skolyoz

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ABSTRACT

Congenital scoliosis is the deformity of the spine that is caused due to intrauterine development anomaly of spine. The defects developing during formation and segmentation periods of the spine may cause a vertebra anomaly. These defects of the vertebra may cause a serious deformity and pulmonary problems because of restriction of thorax development in early childhood. Diagnosis and treatment of congenital scoliosis is important because of its serious complications. We discuss the diagnosis and treatment methods of congenital scoliosis.

KEYWORDS: Classification, Congenital scoliosis, Surgical treatment, Scoliosis

ÖZ

Konjenital skolyoz omurganın intrauterin gelişim anomalisine bağlı gelişen deformitedir. Omurganın formasyonu veya segmentasyonu aşamasında oluşan defektler doğumda ortaya çıkan vertebra anomalilerine yol açar. Vertebralarda oluşan bu defektler erken çocukluk dönemde ciddi deformiteye ve toraks gelişimini engelleyerek ciddi akciğer problemlerine yol açabilir. Bu yüzden erken tanısı ve tedavisi önemlidir. Bu yazıda konjenital skolyozun tanı ve tedavi yöntemleri anlatılacaktır.

ANAHTAR SÖZCÜKLER: Cerrahi tedavi, Konjenital skolyoz, Sınıflama, Skolyoz

INTRODUCTION

Congenital scoliosis is a coronal plane deformity of the spine due to anomalies developing at the intrauterine period. Anomalies developing at the intrauterine period may be segmentation and formation anomaly or both. The prevalence of congenital scoliosis is 1/1000 live births (23). This number has been determined mainly by plain radiographs of lungs that do not show the lumbosacral region so the actual number may be higher.

The growth anomaly of the spine occurs usually in the first 6 weeks of intrauterine life. The configuration of the spine begins at the third or fifth week of intrauterine life with development of somites. This process including the formation and segmentation of the spine is called somatogenesis. The formation phase is completed between 20-30 days. The segmentation phase is completed at the 6th week (3,8).

Etiology

Genetic and environmental factors are the main factors claimed for vertebral development anomalies causing congenital scoliosis. The main environmental factor is hypoxia. Carbonmonoxide like gases that the mother is exposed to are claimed to be responsible. In animal experiments, the negative effect of hypoxia especially in the first 9 days has been demonstrated (7,21). Diabetes mellitus and antiepileptic drugs during gestation are the other risk factors. Although genetic transition is demonstrated in some congenital vertebra anomalies, it is not possible to say that genetic transition definitely exists.

Accompanying Anomalies

Congenital anomalies of different systems and organs may accompany congenital scoliosis. These anomalies (VACTERL) are vertebral, anorectal, cardiac, tracheal, esophageal, renal and extremity anomalies (V: vertebral, A: anorectal, C: cardiac, T: tracheal, E: esophageal, R: renal, L: limbs). Anomalies of neurological tissues are frequently seen through the vertebral axis. Approximately 30-35% of these patients have tethered cord, diplomyelia, Chiari malformation or anomalies of neural tissue (4,22). Congenital cardiac disease (20%), genital and urinary anomalies (20%), muscle and skeleton system anomalies are the other accompanying anomalies (11).

Planning the treatment of these patients and evaluation of other systems are important as we frequently see other system and neural tissue anomalies accompanying congenital scoliosis during the follow up period.

CLASSIFICATION

Although congenital scoliosis is due to segmentation and formation anomalies; most of them are due to a combination of these (18,28).

- Formation defect
    - Partial formation defect

        - Wedge vertebra
    - Complete formation defect
        - Hemivertebra
            - Complete segmented hemivertebra
            - Partial segmented hemivertebra
            - Unsegmented hemivertebra
- Segmentation defect
    - Bloc vertebra
    - Unilateral bar
- Mixed type
    - Combination of segmentation and formation anomalies

Partial formation anomaly is called wedge vertebra (Figure 1A). Also one side is hypoplastic; both pedicles are seen in the wedge vertebra. Complete formation anomaly is described as hemi vertebra and there is unilateral pedicle formation. Hemi vertebra can be classified with the absence or presence of fusion with upper and lower vertebral body, in other words the absence or presence of intervertebral discs between upper and lower vertebra. If intervertebral discs are present between both lower and upper vertebras the condition is called complete segmented hemivertebra (Figure 1B); if intervertebral discs are present between only upper or lower vertebra the condition is called partial segmented hemivertebra (Figure 1C). If there is no intervertebral disc between upper and lower vertebras, in other words if there is fusion, the condition is called unsegmented hemivertebra (Figure 1D).

Segmentation anomaly is abnormal fusion between two vertebras. If this fusion is bilateral it is called bloc vertebra; if it is unilateral it is called unilateral bar.

Mixed anomaly includes both segmentation and formation anomalies and is a frequent pathology. It is hard to describe because of the complex anatomy and mainly causes serious deformities.

Although this classification according to a vertebraal growth anomaly is comprehensible, it has some deficits in evaluation of complex deformities because this method mainly depends on plain radiographs. Some other classification methods are recommended with common usage of 3D tomography and imaging making all pathologies more clear (14).

NATURAL COURSE

To decide the best treatment method, it is important to know the natural course of complex deformities like congenital scoliosis. Statistically, 25% of these congenital curves do not increase, 25% slightly increase, and 50% rapidly increase (20,29). Progression of scoliosis depends on the type of vertebral anomaly and its location. Progression of the curve depends on unbalanced growth on both sides. The age of the patient is therefore important because of the growth potential.

The presence of an intervertebral disc between the vertebral anomaly and adjacent vertebras is the sign of a growing plate in these areas. Asymmetric located growing plates cause

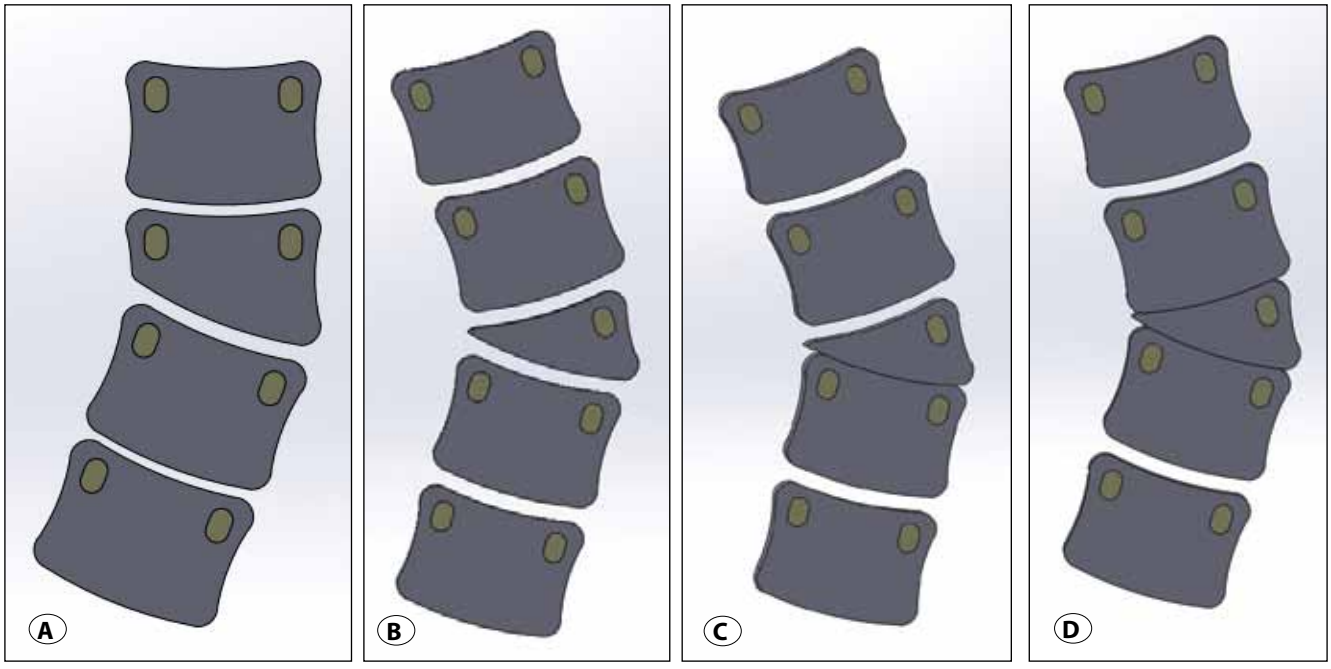

Figure 1: A) Wedge vertebra, **B)** Complete segmented hemivertebra, **C)** Partial segmented hemivertebra, **D)** Unsegmented hemivertebra.

asymmetric growing. Complete segmented hemivertebra therefore has more risk for progression of deformity than unsegmented hemivertebra, as there is a growing plate between hemivertebra and upper and lower vertebras.

Partial segmented hemivertebra anomaly has intervertebral disc and growing area only between the upper and lower side and it therefore has less progression risk than complete segmented hemivertebra but more progression risk than unsegmented hemivertebra. Deformities with a segmentation anomaly due to a unilateral bar have higher progression risk because the contralateral side has growing potential. Bloc vertebra characterised with bilateral fusion has almost no deformity development risk.

Co-occurrence of unilateral bar and complete segmented hemivertebra is rare but it is the anomaly which has the highest risk of deformity development and progression of deformity (18,20).

Cervicothoracal and lumbosacral areas are junctional places and therefore have the potential of sharp angled deformity development. The rapid growing periods are the first 5 years after birth and the growth spurts during adolescence. In these periods growth of the spine is rapid and the possibility of progression of deformity is high. If the type and localization of the anomaly and the age of the patient are considered in congenital scoliosis, more reliable comments can be made about the natural course of the deformity and the best treatment method can be determined (Figure 2A-C).

EVALUATION OF THE PATIENT

Congenital scoliosis is a deformity that occurs in the first years of life. It is rare to see scoliosis in children under 3 years because development of the spine is not completed. However, signs of some accompanying anomalies in the neural axis may be seen in the newborn. Because of its frequent co-occurrence with spinal dysraphism, children with spinal dysraphism signs must be evaluated for a vertebral anomaly. A vertebra anomaly may also be suspected with ultrasonography during routine intrauterine controls. Routine health controls of the newborn or plain radiographs for other reasons can also help to see congenital anomalies in the spine. Intimate follow up of these patients who are diagnosed before development of significant scoliosis is important.

During evaluation of the patient with a diagnosis of vertebra anomaly, it is important to determine the risk of progression of deformity and follow-up periods. All systems of the patients must be evaluated in detail and the data must be noted properly. As in all deformity patients the physical examination, neurological examination and radiological examination is are. In addition, all these patients must be evaluated in detail regarding family history and mother's pregnancy history.

Physical Examination

Physical examination of these patients begins with the assessment of weight and height. Evaluation of the physical development with age may give information about the growth potential of the patients. Skin signs of spinal dysraphism like

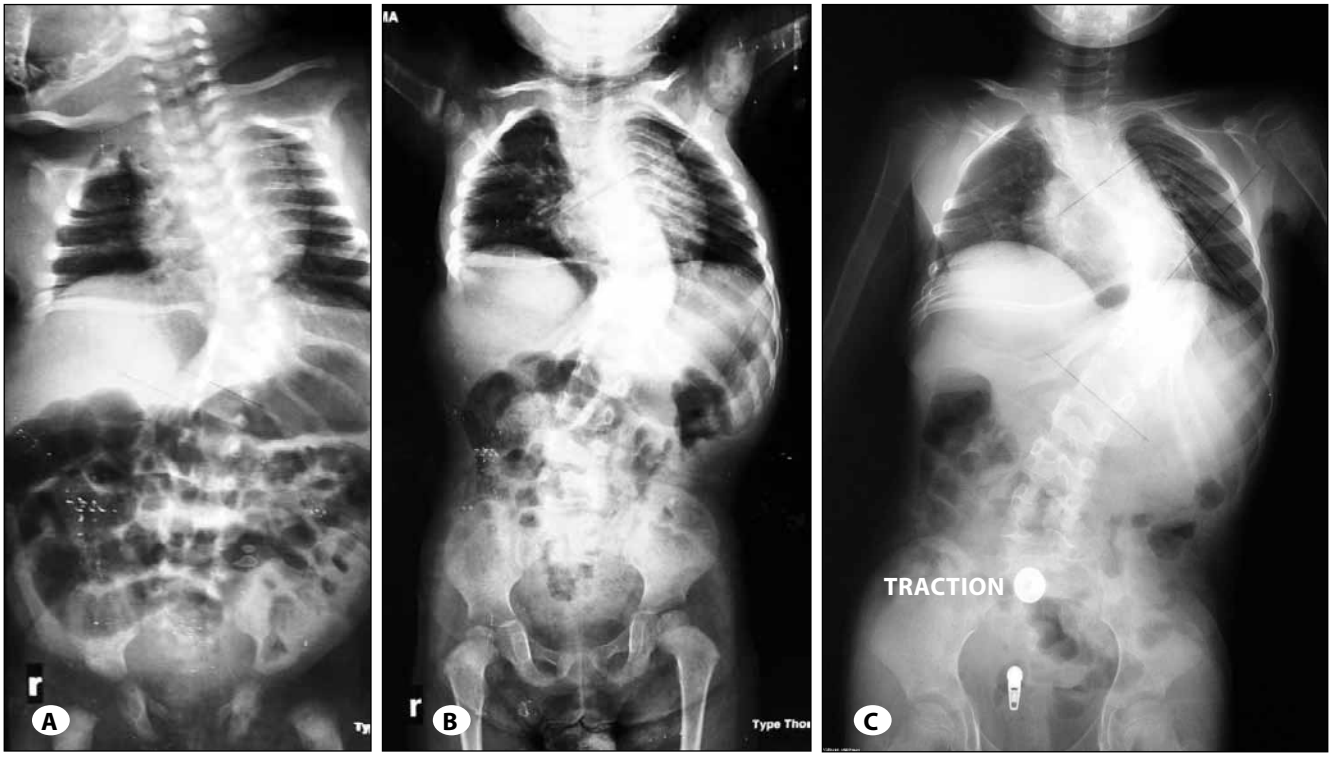

Figure 2: T10 complete segmented hemivertebra diagnosed at the newborn period **A)** not treated, **B)** 2 years **C)** and 6 years later showing the progression of deformity.

hirsutism, dermal sinus, change of color are checked. The lower extremities are examined for intraspinal pathologies or accompanying skeletal system anomalies. The presence of a chest anomaly is important. It must be kept in mind that these patients have the potential of developing restrictive pulmonary disease due to underdevelopment of the chest. Coronal and sagittal spinal balance, pelvis and shoulder asymmetry are also evaluated.

Neurological Examination

Neurological examination is important because anomalies like tethered cord syndrome can be seen in these patients. Neurological examination must be done in detail for all patients and children must be evaluated for the development of motor function and sphincter control.

Radiological Examination

We can classify the diagnostic procedures as necessary or selective in order to diagnose, follow up and treat the deformity. Anterior-posterior (AP) and lateral radiographs including the whole spine are necessary procedures. As in all other scoliosis patients, plain radiographs including the pelvis and cervical region to evaluate sagittal and coronal balance are necessary (Figure 3). Lateral bending graphies in the supine position or forced prone graphies by pressing on the apex of the curve may be obtained to determine the flexibility of the deformity. Congenital scoliosis is usually rigid but compensatory curves developing due to the main curve may be flexible. This is important when determining the sufficiency of brace treatment as a treatment method.

Plain scoliosis graphies are generally sufficient for follow up. However, 3D tomography may be helpful to determine the complex structure of the congenital scoliosis deformity (Figure 4).

The time of magnetic resonance imaging (MRI) is controversial. MRI is definitely indicated for patients where spinal dysraphism is suspected. However, it must be kept in mind that intradural pathologies that do not have spinal findings such as the Chiari malformation and tethered cord syndrome may accompany vertebra anomalies. Children usually need to be sedated during MRI. This is a limiting factor for MRI but MRI must be performed if there is a little doubt about intradural pathology.

Chest cage anomalies are frequently seen in congenital scoliosis. With PA radiographs costa anomalies and pulmonary development may be evaluated. Patients with scoliosis developing under 5 years may have serious distortion of pulmonary capacity due to restricted chest cage and these children must be closely followed up for pulmonary capacity (16).

TREATMENT

The aim of treatment is early diagnosis and early surgery if needed. The main objectives can be listed as follows (16,25):

– Correction if deformity has developed
– Prevention of deformity development if deformity is absent
– Maintaining the growth potential of spine and ensuring its growth
– Ensuring the development of the chest cage and maintaining pulmonary functions

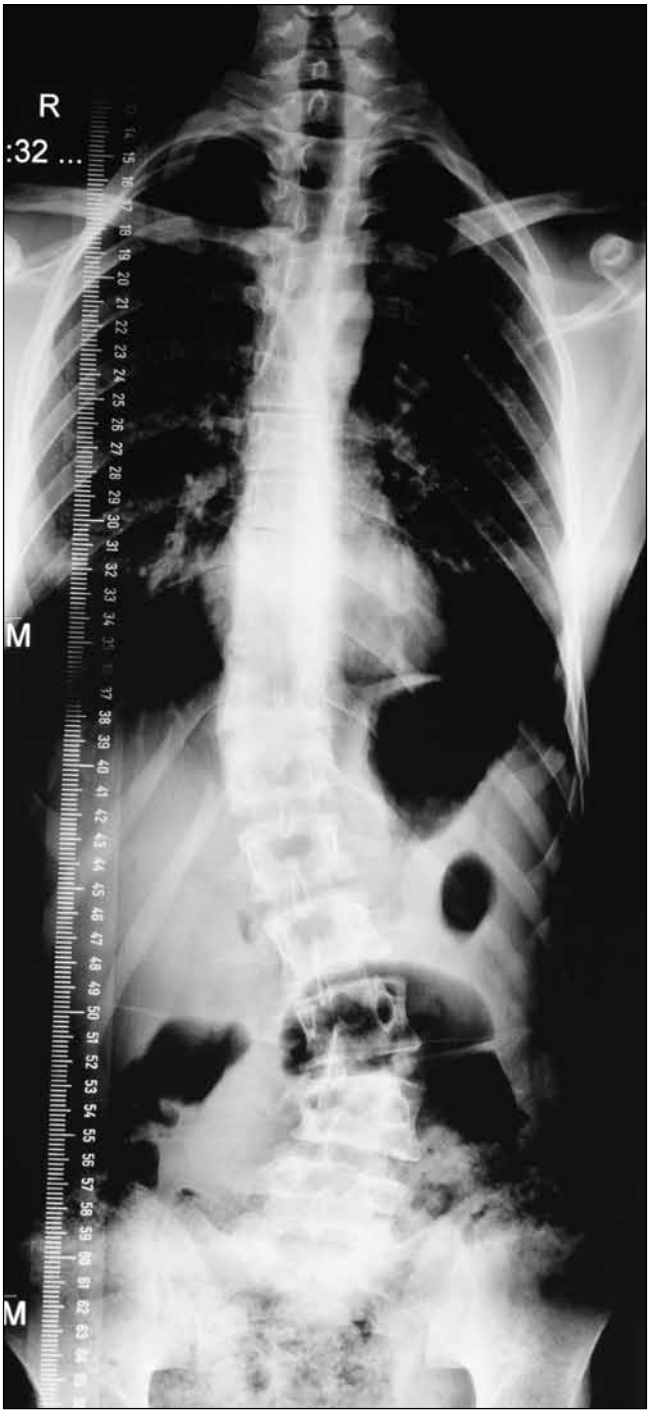

Figure 3: Scoliosis graph including the whole cervical and pelvic region.

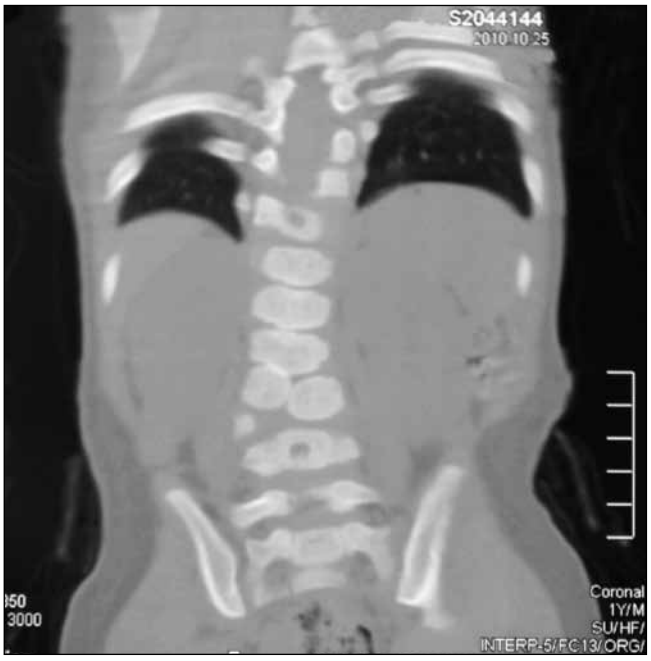

Figure 4: Multislice coronal reconstruction with tomography to evaluate the deformity better.

When a congenital vertebra anomaly is determined, its type, location of anomaly and the age of the patient is important to decide about the treatment method. Maximum growth period of the spine is between 0-5 and 10-15 years (2). Development of a deformity must be frequently followed up especially at the rapid growth period (0-5 years and 10 years to puberty). Follow up intervals must be maximum 4-6 months during this period. The treatment method must be decided according to the increasing potential of the deformity.

Conservative Treatment

The benefit of treatment with a brace has been demonstrated in patients with a long segment curve and a curve that is flexible as shown with supine lateral bending radiographs. Treatment with a brace may also be helpful for compensatory curves. Generally, brace treatment is not used for congenital scoliosis because the segment is mainly short and rigid.

Surgical Treatment

Surgical treatment is the only choice for patients with high progression potential and progression during follow up. Surgical treatment is the general treatment method as the angle of curve increases in 75% of the patients and treatment with a brace is not sufficient.

Surgical Treatment Options

Surgical treatment in congenital scoliosis is performed for two reasons: to correct the deformity or to prevent deformity development. The deformity is usually rigid in congenital scoliosis. Surgery performed to correct the developed deformity are therefore difficult and the complication rate is relatively high. Surgical treatment options are performed to prevent the development of deformity before the patient's development is completed or before compensatory curves occur is more valuable for congenital scoliosis. Surgical treatment options can be listed as follows:

- Correction with instrumentation and fusion
- In situ fusion (with or without instrumentation)
- Combined anterior and posterior fusion
- Convex hemiepiphysisodesis
- Excision of hemivertebra
- Growth-oriented treatment (growing rod)
- VEPTR (vertical expandable prosthetic titanium rib)

Correction with Instrumentation and Fusion

This is mainly performed to correct deformity. The aim is to provide spinal balance and correct the deformity. Pedicle screws and, depending on the location, lamina or pedicle hooks are reliable fixation instruments for congenital deformity surgery. Correction can be achieved with applying a cantilever, derotation, compression or distraction forces to the spine by rods (10,24). Congenital scoliosis cases are mainly rigid deformities and maneuvers without releasing the spine cannot correct the curves. Procedures to release the spine such as osteotomy, release of anterior longitudinal ligament with anterior procedure, discectomy, and excision of facet joints must therefore be performed for rigid deformities and especially for curves over 70° (5). Anterior procedure may be performed not only for discectomy or to release anterior longitudinal ligament but to apply the instrument that will support the anterior column in some patients. Anterior fusion must be performed especially for patients with long segment stabilization and fusion to prevent crank shaft phenomenon who are at a growing age (15).

It must be kept in mind that long segment fusion for children at the growing period will negatively effect the growth of the spine and total body. Surgical planning must therefore be used to keep the fusion level with minimum segment (Figure 5a, 5b, and 5c).

In Situ Fusion (Fusion with or without Instrumentation)

This surgical procedure is performed to prevent the development of deformity. Fusion surgery is the most reliable and effective surgery for congenital scoliosis. It can be performed for patients whose deformities are determined at an early period. Curves of a maximum 50 degrees, with high increase potential of deformities like complete segmented hemivertebra, unilateral bar or both of these pathologies are ideal candidates for this surgery (12,19). Anterior procedure may be added for patients with potential of growth in the anterior spine. Posterior fusion can also be performed without instrumentation but short segment instrumentation can be used to increase the fusion rate and to prevent necessity of corset fusion (Figure 6A,B).

Convex Hemiepiphysisodesis

This procedure provides correction of deformities as the patient grows up (27). Patients with incomplete deformity development, complete segmented hemivertebrae and with potential of growth on the concave side of the deformity are ideal candidates for this procedure (Figure 7). Fusion is performed on the convex side of the deformity. The growth potential of the concave side provides correction of

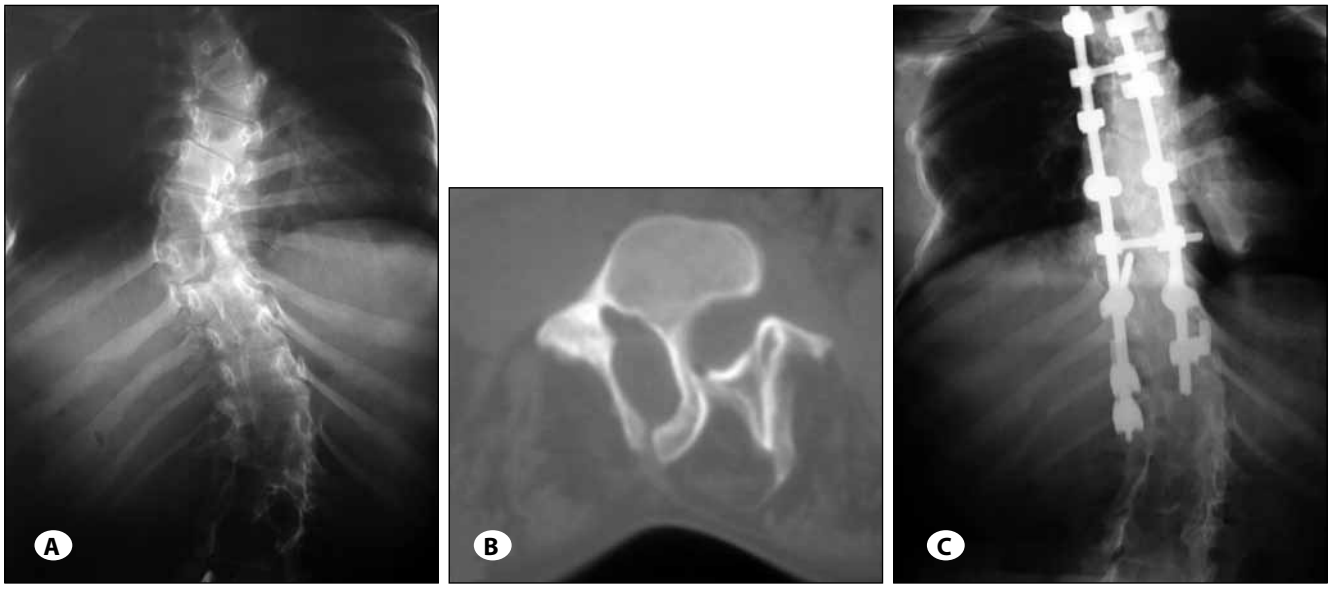

Figure 5A-C: A 15-year old patient with rigid deformity and diastematomyelic made correction in same surgery.

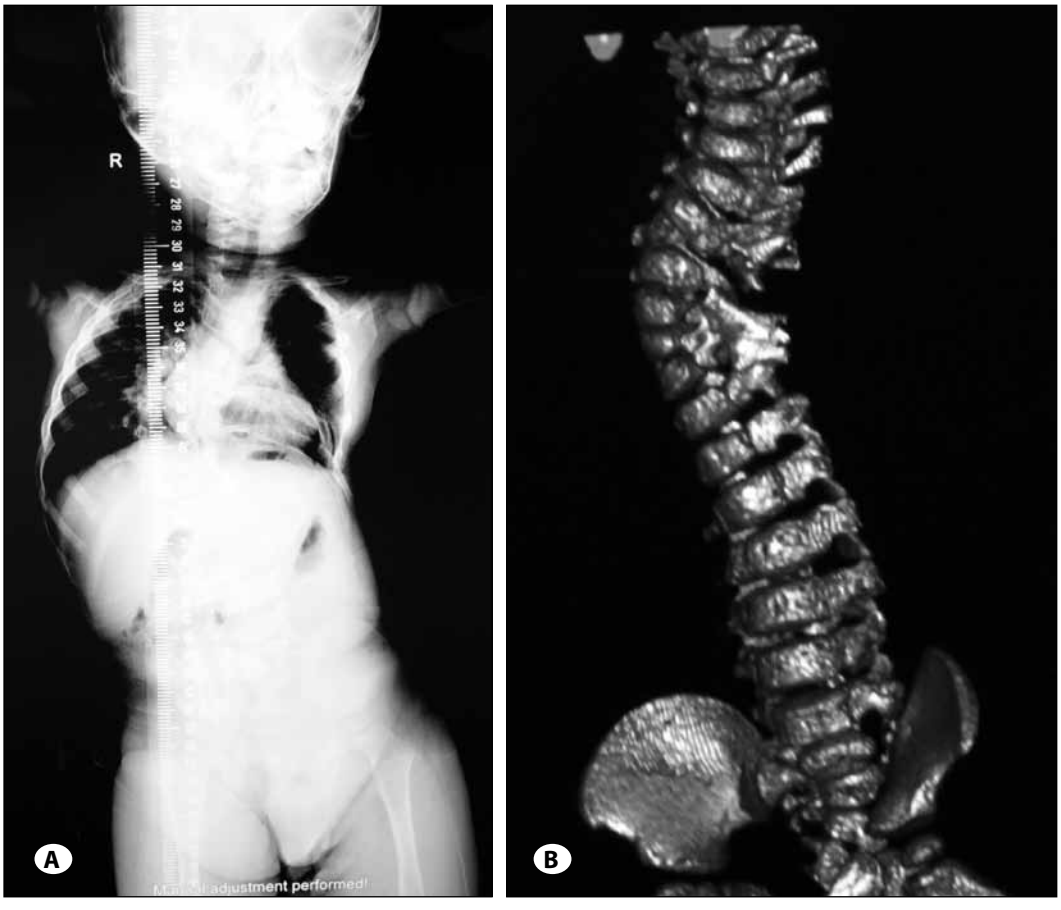

Figure 6 A, B: Ideal in situ fusion case for preventing increasing deformity in a 3-year-old patient with a unilateral bar.

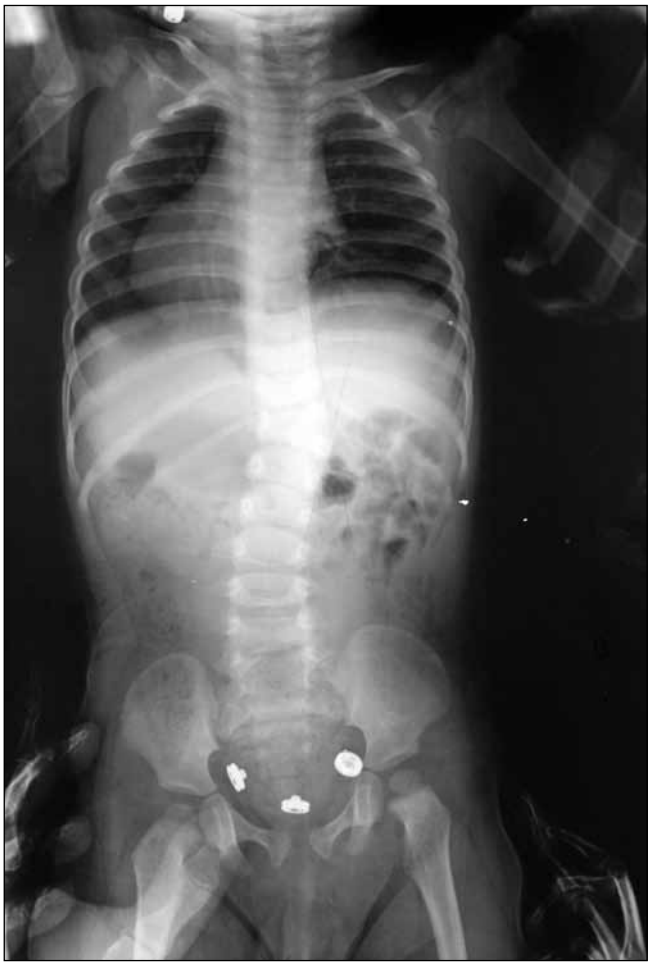

Figure 7: L1 complete segmented hemivertebra, convex hemiepiphysisodesis case.

deformity in time. The correction rate of the deformity cannot be predicted. Performing combined anterior or posterior procedure enables controlling deformity in both the coronal and sagittal planes (9).

Excision of Hemivertebra

This is a procedure performed to correct deformity. Patients under 5 years with isolated partial or complete segmented hemivertebrae located at the thoracolumbar or lumbar region are ideal candidates (12). This procedure may also be performed at an older age and with a deformity which has completed development.

This procedure can be performed only posteriorly or with combination of anterior and posterior procedures (13). Correction can be obtained with pedicle screws or hooks instrumented on the convex side after excision of hemivertebra. Pedicle screws or hooks can be instrumented on the concave side for stabilization.

Hemivertebra is a certain surgical method. It provides correction of deformity immediately and protection of growth potential because of short segment fusion.

Growth-Oriented Treatment (Applying Growing Rod)

Fusion is the golden standard for deformity surgery but performing fusion at the growing age causes the spine and total body to be short. In some cases fusion negatively affects chest cage development and causes complications like lung failure. Short segment fusion or treatment without fusion are therefore ideal methods, especially for patients at the growing age (1).

The aim is correction of deformity with ensuring growth of the spine. Patients under 5 years and with a deformity affecting

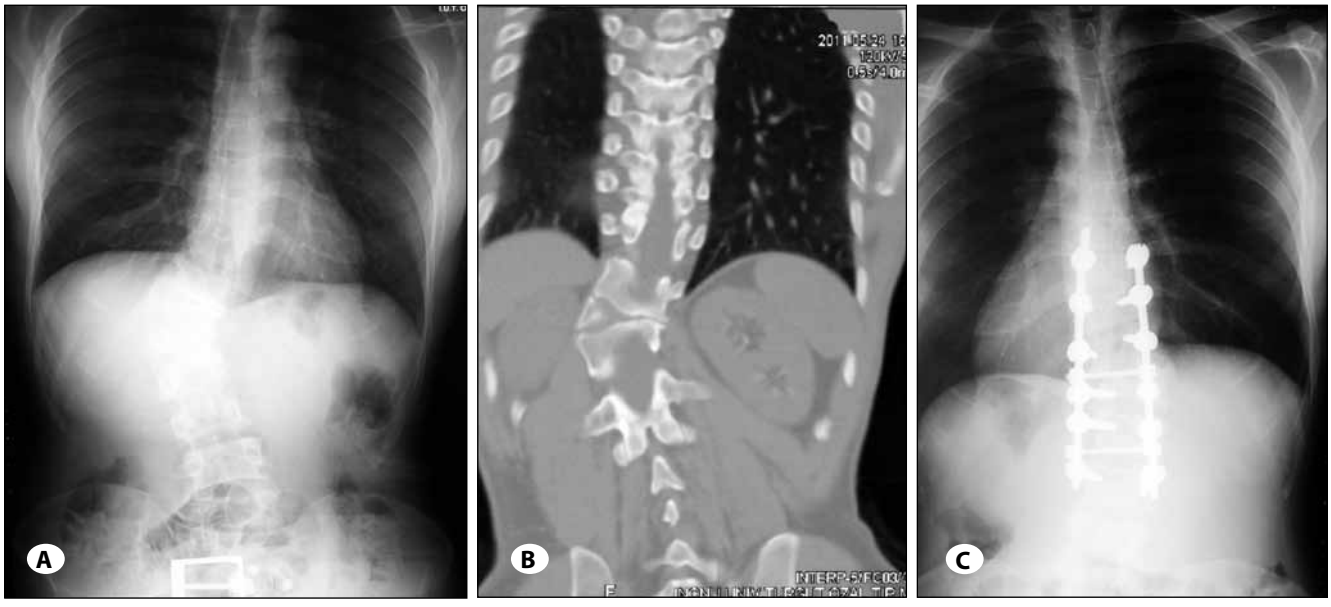

Figure 8A-C: A15-year-old patient development of deformity without fusion.

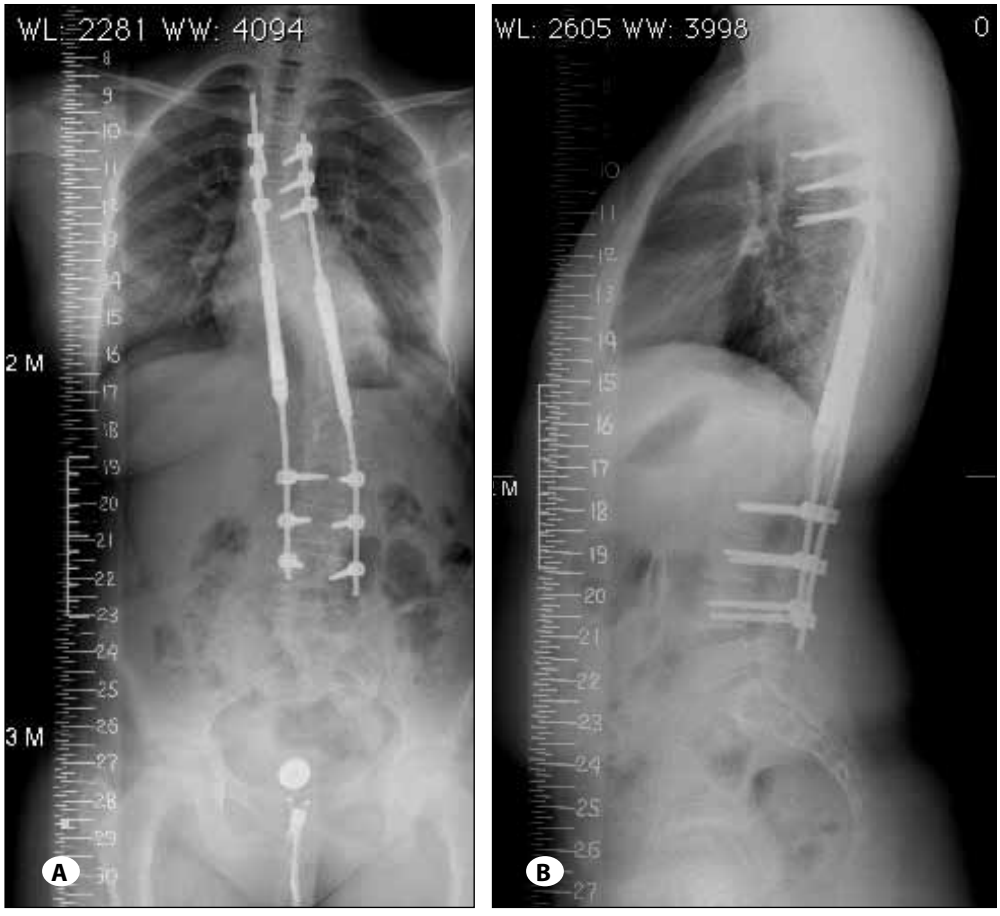

Figure 9A, B: Growing rod application for an 11- year-old patient to prevent progression of deformity.

the long segment of the spine are ideal candidates. Pedicle screws or hooks are inserted to the proximal or distal ends of the deformity. Distraction is performed with a rod. Distraction is obtained with the increase of the degree of the curve. In this way, prevention of deformity development is obtained while providing growing of the spine.

This procedure is mainly performed with repeated surgeries by extending the rod. Patients frequently need operation with this procedure this causes medical problems, dermal problems and increased infection rate. Today this problem is somewhat controlled with external and magnetic controlled growing rod technologies (Figure 9a and 9b).

VEPTR (Vertical Expandable Prosthetic Titanium Rib)

This is applying instrument to the costa for distraction. It is the surgical procedure performed for patients at a growing age and for a deformity affecting the thorax to increase thoracic capacity. It is performed by opening the costa fusioned on the concave side of thorax, and the thoracal volume is expanded by distraction in 4-6 month intervals (6). After sufficient thoracal volume is obtained at growing age, fusion surgery is performed.

REFERENCES

1. Akbarnia BA, Marks DS, Boachie-Adjei O: Dual growing rod technique for the treatment of progressive early-onset scoliosis. Spine 30: 46 – 57, 2005
2. Akbarnia BA: Management themes in early onset scoliosis. J Bone Joint Surg Am 89: 42-54, 2007
3. Arlet V, Odent T, Aebi M: Congenital scoliosis. Eur Spine J 12: 456-463, 2003
4. Belmont PJ Jr, Kuklo TR, Taylor KF, Freedman BA, Prahinski JR, Kruse RW: Intraspinal anomalies sassociated with isolated congenital hemivertebra, the role of routine magnetic resonance imaging. J Bone Joint Surg 86: 1704–1710, 2004
5. Bridwell KH: Decision making regarding Smith-Petersen vs. pedicle subtraction osteotomy vs. vertebral column resection for spinal deformity. Spine 31: 171–178, 2006
6. Campbell RM Jr, Smith MD, Hell-Vocke AK: Expansion thoracoplasty. The surgical technique for opening-wedge thoracostomy. Surgical technique. J Bone Joint Surg Am 86: 51-64, 2004
7. Farley AF: Etiology of congenital scoliosis. Semin Spine Surg 22: 110-112, 2010
8. Ganey TM, Ogden JA: Development and maturation of the axial skeleton, Weinstein SL (ed), The Pediatric Spine ikinci baskı, Philadelphia: Lippincott, Williams and Wilkins, 2001:3-54

9. Ginsburg G, Mulconrey DS, Browdy J: Transpedicular hemiepiphysiodesis and posterior instrumentation as a treatment for congenital scoliosis. J Pediatr Orthop 27: 387-391, 2007
10. Hedequist DJ, Hall JE, Emans JB: The safety and efficacy of spinal instrumentation in children with congenital spine deformities. Spine 29: 2081–2087, 2004
11. Hedequist D, Emans J: Congenital scoliosis: A review and update. J Pediatr Orthop 27: 106-116, 2007
12. Hedequist DJ: Surgical treatment of congenital scoliosis. Orthop Clin N Am 38: 497-509, 2007
13. Jalanko T, Rintala R, Puisto V, Helenius I: Hemivertebra resection for congenital scoliosis in young children, comparison of clinical, radiographic, and health-related quality of life outcomes between the anteroposterior and posterolateral approaches. Spine 36: 41-49, 2011
14. 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 calssification for progressive vertebral anomalies requiring operetive treatment. Spine 34:1756-1765, 2009
15. Kesling KL, Lonstein JE, Denis F, Perra JH, Schwender JD, Transfeldt EE, Winter RB: The crankshaft phenomenon after posterior spinal arthrodesis for congenital scoliosis. A review of 54 patients. Spine 28: 267–271, 2003
16. Kose N, Campbell RM: Congenital scoliosis. Med Sci Monit10: 104-110, 2004
17. Maienbacher MK, Han JS, O'Brien ML, Tracy MR, Erol B, Schaffer AA, Dormans JP, Zackai EH, Kusumi K: Moleculer analysis of congenital scoliosis. A candidate gene approach. Hum Genet 116: 416-419, 2005
18. Mc Master MJ, Ohtsuka K: The natural history of congenital scoliosis. A study of two hundred and fifty-one patients. J Bone Joint Surg Am 64: 1128–1147, 1982
19. Mc Master MJ, David CV: Hemivertebra as a cause of scoliosis. A study of 104 patients. Surg Br 68: 588-595, 1986
20. Mc Master MJ: Congenital scoliosis caused by a unilateral failure of vertebral segmentation with contralateral hemivertebrae. Spine 23: 998–1005, 1998
21. Murakami U, Kameyama Y: Vertebral malformations in the Mouse fetus caused by maternal hyposia during early stages of pregnancy. J Embryol Exp Morphol 11: 107-118, 1963
22. Prahinski JR, Polly DW Jr, McHale KA, Ellenbogen RG: Occult intraspinal anomalies in congenital scoliosis. J Pediatr Orthop 20: 59-63, 2000
23. Shands AR Jr, Bundens WD: Congenital deformities of the spine. an analysis of the roentgenograms of 700 children. Bull Hosp Jt Dis 17: 110-133, 1956
24. Suk SI, Kim WJ, Lee SM, Kim JH, Chung ER: Thoracic pedicle screw fixation in spinal deformities. Are they really safe? Spine 26: 2049–2057, 2001
25. Thompson GH, Lenke LG, Akbarnia BA, McCarthy RE, Campbell RM Jr: Early onset scoliosis, future directions. J Bone Joint Surg Am 89: 163-166, 2007
26. Thompson G, Akbarnia BA, Campbell RM: Growing rod techniques in early-onset scoliosis. J Pediatr Orthop 27: 354-361, 2007
27. Uzumcugil A, Cil A, Yazıcı M, Acaroglu E, Alanay A, Aksoy C, Surat A: Convex growth arrest in the treatment of congenital spinal deformities, revisited. J Pediatr Orthop 24:658-666, 2004
28. Winter RB: Congenital deformities of the spine. New York, NY: Thieme-Stratton, 1983
29. Winter RB, Lonstein JE, Boachie-Adjei O: Congenital spine deformity. J Bone Joint Surg Am 78: 300– 311, 1996