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Report of Two Adult with Occult Cervical Spinal Dysraphism and Scoliosis

Gizli Spinal Disrafizm ve Skolyozlu İki Erişkin Olgu

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Özet

Spinal disrafizm (SD), embriyogenez sırasında nöral tüpün kapanmasındaki bozukluğa bağlı omurganın kemik ve nöral yapılarının kısmi birleşmesi veya malformasyonudur. Omurganın arka kısmının orta hat boyunca tam olarak kapanmaması söz konusudur. SD sıklıkla torakolomber ve ve lumbosakral seviyelerde oluşur ve servikal SD tüm spinal anomalilerin %1-5'ini oluşturan nadir bir durumdur. SD açık ve kapalı(gizli) olmak üzere iki şekilde ortaya çıkmaktadır. Gizli SD, genellikle düz grafi veya manyetik rezonans görüntüleme sırasında tesadüfen saptanmaktadır. Klinik olarak önemli gizli SD, kas kuvvetsizliği, cilt anormalliği, Klippel-Feil Sendromu, torasik hemivertebra, ayak deformitesi, duysal anormallik, üriner inkontinans, yürüyüş bozukluğu ve skolyoza neden olabilir. Boyun ve sırt ağrısıyla başvuran ve gizli SD ve torakolomber skolyoz saptanan iki erişkin kadın olguyu sunuyoruz. Servikal SD nadiren bildirilmesine rağmen, dermatolojik, nörolojik ve ürogenital hastalıklarla birlikte skolyoz gibi ortopedik bozukluklarla da birlikte ortaya çıkabilir.

Anahtar Kelimeler: Torakolomber Skolyoz, Spinal disrafizm.

Abstract

Spinal dysraphism (SD) is the partial fusion or malformation of bone and neural structures of the spine by errors in the closure of the neural tube during embryogenesis. SD commonly occurs at the thoracolumbar and lumbosacral levels, and cervical SD is an uncommon condition that comprises 1–5% of all spinal anomalies. Occult SD is generally diagnosed incidentally on plain radiography or magnetic resonance imaging. Clinically important occult SD may cause muscle weakness, cutaneous abnormality, Klippel-Feil syndrome, thoracic hemivertebra, foot deformity, sensory abnormality, urinary incontinence, gait abnormality and scoliosis. We report two adult females with occult cervical spinal dysraphism and thoracolumbar scoliosis who presented with neck and shoulder pain. Although cervical SD has rarely been reported, this report highlights that may be associated with orthopedic abnormalities such as scoliosis as well as cutaneous, neurologic and urogenital disorders.

Keywords: Thoracolumbar scoliosis, spinal dysraphism.

Introduction

Spinal dysraphism (SD) is the incomplete fusion or malformation of bone and neural structures of the spine region by errors in the closure of the neural tube during the fourth week of embryogenesis. The incidence of these defects is from 0.5 to 5 cases per 1000 births, with significant geographical variation (1).

Occult SD is rarely linked with complications or symptoms. Occult SD is generally diagnosed incidentally on plain radiography or magnetic resonance imaging (MRI) (2).

SD commonly occurs at the thoracolumbar and lumbosacral levels, and cervical SD is a very rare condition that comprises 1–5% of all spinal anomalies (2).

We report two adult females with occult cervical SD and scoliosis who had presented with neck and shoulder pain.

Case Report 1

A 21-yr-old woman admitted to an outpatient physical therapy clinic with a 2-yr history of neck and right shoulder pain that was insidious at onset. Her pain was improved with rest, heat application and medical treatment. On physical examination, she had active myofascial trigger points in both upper trapezius muscles. Her cervical active range of motion was within functional limits and painless in all planes. She had drooping shoulder on the left side. Adam's forward bending test was positive. A cervical and thoracic spine x-ray demonstrated defective fusion of posterior bony elements in lower cervical and upper thoracic vertebrae and thoracolumbar scoliosis with a 15° curve angle (Fig. 1A and 1B).

Axial T2 weighted MRI imaging of C5 and C6 vertebra showed deficient posterior lamina and defective fusion of posterior elements (Fig. 2).

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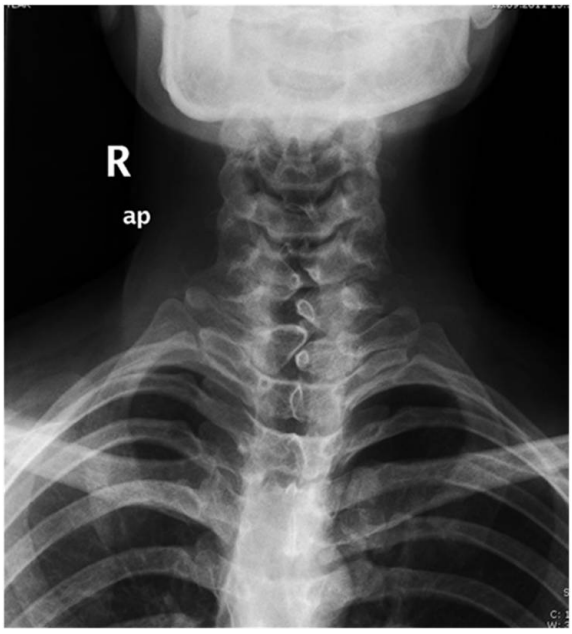

Figure 1A. Cervical radiography shows defective fusion of posterior bony elements at C6, C7 and T1.

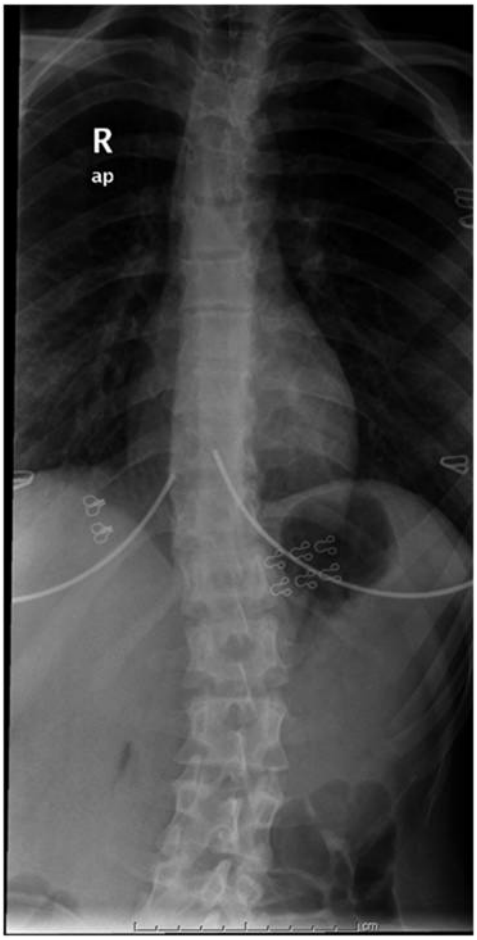

Figure 1B. Thoracic radiography shows thoracolumbar scoliosis with an S-form curve.

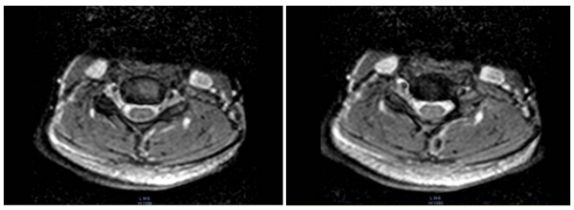

Figure 2. Axial T2 weighted MRI imaging of C5 and C6 vertebra showing deficient posterior lamina and defective fusion of posterior elements.

Case Report 2

A 37-yr-old woman admitted to an outpatient physical therapy clinic with a 4-yr history of neck pain. Her pain was increased with neck movements, especially in the anteflexion. She had active myofascial trigger points in left upper trapezius muscle. Her cervical active range of motion was within functional limits in all planes but slightly painful. Neurologic examination was normal. A cervical and thoracic spine x-ray demonstrated defective fusion of posterior bony elements in cervical fifth vertebrae and thoracolumbar scoliosis with a 30° curve angle (Fig. 3A and 3B).

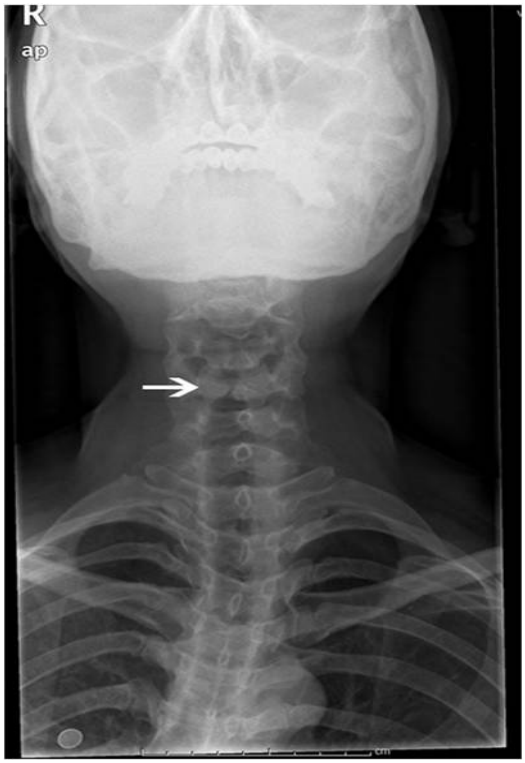

Figure 3A. Cervical radiography shows defective fusion of posterior bony elements at C5.

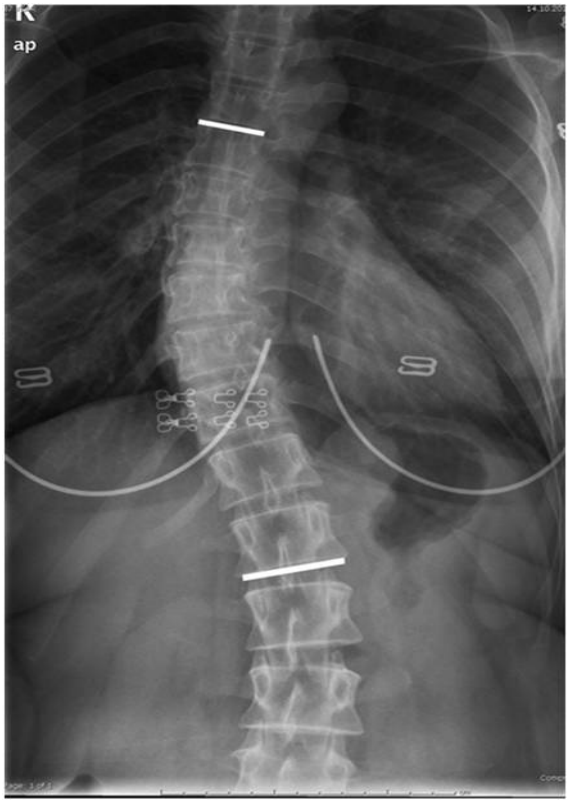

Figure 3B. Thoracic radiography shows thoracolumbar scoliosis with an S-form curve. The apex of the major curve is located at T9, with a curve length of 8 vertebrae.

Discussion

The term “spina bifida” is still commonly used as a synonym of SD, although it properly refers to defective fusion of posterior spinal bony elements. The terms “spina bifida aperta” or “cystica” and “spina bifida occulta” were once used to refer to open spinal dysraphism and closed spinal dysraphism, respectively, but are no longer widely used (3).

The most frequent etiologies of SD include a low socioeconomic level, inadequate folic acid intake, oral contraceptives, ovulation stimulants, diabetes mellitus, zinc deficiency, and anticonvulsants (especially valproic acid and carbamazepine), obesity, vitamin B12 deficiency, high body temperature, metabolic teratogenic agents, familial history of defects in the neural tube, and environmental contamination (2-4). Familial history is positive in approximately 10% of cases; if the couple already has a child with SD, the chance of reoccurrence of the disease is from 3.5% to 5.5% (5).
The neonate with occult SD may not present clinical manifestation, but may be associated to cutaneous stigmas that indicate dysraphism: lipomas, abnormal hair, dimples, skin tags and alterations in gluteal fold. The diagnosis of these lesions is difficult because there is no clinical manifestation in most patients. Therefore they may be overlooked by the initial pediatric evaluation. Ultrasonography and magnetic resonance imaging are the chosen methods for the screening and diagnostic confirmation of occult SD (1).

Clinically important occult SD may cause muscle weakness, cutaneous abnormality, Klippel-Feil syndrome, thoracic hemivertebra, foot deformity, sensory abnormality, urinary incontinence, gait abnormality and scoliosis (4).
Our two patients presented with neck pain and they showed cervical SD and thoracolumbar scoliosis on the radiographies. Although one patient had a SD at the C5 vertebra and other patient showed the three-level SD including C6, C7 and T1 vertebrae.

Although cervical SD has rarely been reported, this report highlights that may be associated with orthopedic abnormalities such as scoliosis as well as cutaneous, neurologic and urogenital disorders.

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