CASE REPORT

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A Rare Cause of Scoliosis: Brachyolmia Type 3

Skolyozun Nadir Bir Nedeni: Brakiyolmia Tip 3

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ABSTRACT

Brachiolmia type 3 is a rare genetic syndrome that can cause curvature of the spine, kyphosis, and chronic spine pain. In this case report, skeletal dysplasia was observed in the anamnesis of the patient who presented with low back pain and was noticed to have scoliosis during the physical examination. Suspicion is based on radiographic findings and definitive diagnosis is made by genetic testing. In treatment, a rehabilitation program should be created to manage the symptoms. Scoliosis exercises, breathing exercises, posture exercises and analgesic electrotherapy currents should be kept in mind in the treatment.

Keywords: *Brachyolmia, rehabilitation, scoliosis*

ÖZET

Brakiyolmia tip 3, omurganın eğriliğine, kifoza ve kronik omurga ağrısına neden olabilen nadir bir genetik sendromdur. Bu olgu sunumunda bel ağrısı şikayeti ile başvuran ve fizik muayenesinde skolyozu fark edilen hastanın anamnezinde iskelet displazisi izlendi. Şüphe, radyografik bulgulara dayanır ve kesin tanı genetik testlerle konur. Tedavide semptomların yönetimine yönelik bir rehabilitasyon programı oluşturulmalıdır. Tedavide skolyoz egzersizleri, nefes egzersizleri, postür egzersizleri ve analjezik elektroterapi akımları akılda tutulmalıdır.

Anahtar Kelimeler: *Brakiyolmia, rehabilitasyon, skolyoz*

INTRODUCTION

Brachyolmia is a rare genetic syndrome and was first defined in 1989 (1). The word brachyolmia was introduced to describe a bone dysplasia which is characterized clinically by short stature and radiographically platyspondyly. Brachyolmia type 3 can present with curvature of the spine, kyphosis and chronic spinal pain. The findings on the plain radiograph are suspected and a definitive diagnosis is made with genetic tests, most commonly identifying mutations in the TRPV4 genes .

TRPV4 is a calcium-permeable cation channel and is expressed in many tissues such as the brain, eyes, kidney tissue, urinary system, gastrointestinal tract, pancreas, skin, epithelial tissue, vascular endothelium, and many musculoskeletal tissues (2). Mechanosensitive TRPV4 plays a critical role in the physiological connection between mechanical loading and extracellular matrix biosynthesis in the growth plate (3). TRPV4 activation causes SOX9 gene expression and endochondral ossification in chondrogenic cells (4). Additionally, TRPV4-induced neuronal death is mediated by enhancing the neuroinflammatory response, and this action plays a role in neuronal injury following status epilepticus. TRPV4 autosomal dominant mutation is associated with skeletal dysplasias such as familial digital arthropathy-brachydactyly, autosomal dominant brachyolmia, spondylometaphyseal dysplasia, Kozlowski and Maroteaux type, parastremmatic dysplasia and metatropic dysplasia. Neuropathies related with this gene are Charcot-Marie-Tooth disease type 2C, scapuloperoneal spinal muscular atrophy and congenital distal spinal muscular atrophy (5).

When encountering scoliosis while investigating a pain complaint, brachiolmia, one of the causes of syndromic scoliosis, should be considered (6).

CASE REPORT

A 39-year-old female patient applied to the physical therapy and rehabilitation clinic with a complaint of back pain. In her history, it was learned that this complaint was

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mechanical in nature. This localized pain has been present for 2 years (chronic) and she had no night pain. There was no numbness or tingling in the arms and legs. There is no history of trauma, epilepsy, arthropathy, or fracture in the patient's medical history. At the same time, it was learned that both the patient and her son had a genetic disease called Brachiolmia type 3 as a result of heterozygous TRPV4 mutation. Having the genetic disease test result in hand provides ease of diagnosis. Her son does not have epilepsy and she does not have any other children. She stated that her son also has back pain but does not have any additional diagnosis. There are no short stature, short extremities, bradydactyly, absence of fingers, disproportion of fingers, chest deformity, history of retinal detachment, history of kidney disease, growth retardation, skeletal deformity, consanguineous marriage in the family. Her menstrual cycles are regular. During physical examination, scapular asymmetry was observed. Paravertebral tenderness and myofascial trigger points around the scapula were detected by palpation. Cervical joint range of motion was open without pain, and lumbar joint lateral flexion and rotation were limited bilaterally. Hand-finger-floor distance was 15 cm, and Adam's forward bending test was positive. Beighton score:0/9. Hip joint range of motion was open. The visual pain scale for the patient was 8. No signs of atrophy or neuropathy were observed. Her height is 155 cm and her weight is 45 kg. Chest expansion and lung auscultation are normal. Other system examinations are normal.

A-P scoliosis radiograph revealed right thoracolumbar scoliosis with a 23.5 degree cobb angle between the T6-L2 end vertebrae. Lateral scoliosis radiography showed hypokyphosis with 5 degree of sagittal Cobb angle, height loss in the vertebral bodies, and platyspondylia (Figure 1).

After the evaluation, the patient's treatment was planned as schroth exercises, breathing exercises, posture exercises and Transcutaneous electrical nerve stimulation (TENS) application to the painful waist area within the scope of scoliosis rehabilitation. After 15 sessions of treatment, the patient's VAS decreased from 8 to 6. The patient was called to the polyclinic for physical examination and x-ray 6 months later.

DISCUSSION

Brachyolmia type 3, caused by TRPV4 (transient receptor potential vanilloid 4) autosomal dominant mutation, may present with flattened vertebrae (platyspondylia), kyphosis, scoliosis, and short stature (7). It is classified as congenital or acquired based on its onset. If platyspondylosis affects all vertebrae, it is called generalized; if it affects several vertebrae, it is called multiple; if it affects one vertebra, it is called localized. Causes of generalized platyspondylia: osteochondrodysplasias, endocrine diseases, metabolic diseases, hematological diseases. In case of localised platyspondylia, histiocytosis, infection, trauma and tumor should be considered (8).

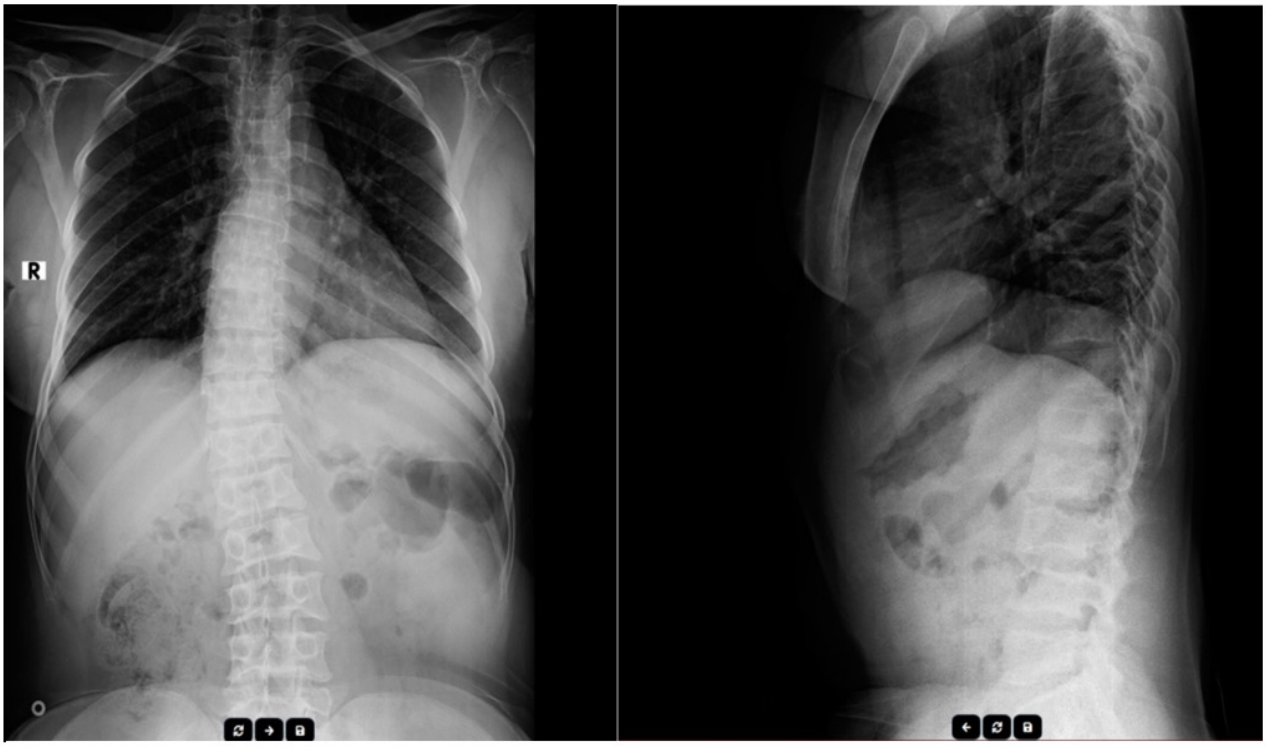


Figure 1. A-P and lateral scoliosis radiograph

A total of 31 cases of autosomal dominant brachyolmia have been reported in case reports; Gardner et al. reported 2 patients (9), Grigelioniene et al. reported 12 patients (10), Lim et al. reported 1 patient (11), Nishimura et al. reported 6 patients (12), and Rock et al. reported 10 patients (13). Unlike our case, femoral lines were observed on radiography in the literature (11). Only Rock et al. evaluated the cases genetically and clinical features were not reported (13).

Brachyolmia type 3 may be asymptomatic or cause complaints such as short stature, stiffness in the spine, spine pain, limb pain, paresthesia, and premature pubarche. Radiological findings may include platyspondylia, kyphosis/scoliosis, decreasing interpeduncular distance towards the lower vertebrae (overfaced pedicles), proximal and distal femoral metaphyseal lines and striations (14).

Treatment should be managed by a multidisciplinary team in collaboration with a neurologist, psychiatrist, orthopedist and physiatrist. Attention should be paid to symptom management (15). In cases of spinal pain or limb pain, analgesic electrical currents should be considered. Schroth therapy can be applied for scoliosis. Posture training should be applied. Considering that pulmonary functions may be affected in case of kyphoscoliosis, breathing exercises including pursed-lip breathing and diaphragmatic breathing should be added to the treatment even in mild cases. If restrictive lung disease, upper cervical spinal instability, or spinal myelopathy develop due to kyphoscoliosis, surgical treatment may be recommended (15). Although there is no specific treatment, the prognosis is good (16). In the controls, attention should be paid to the progression of scoliosis and whether there is any respiratory effect.

Conclusion

Brachyolmia type 3 is a rare skeletal dysplasia that can present with pain. A regression was detected in the patient's VAS score with physical therapy and rehabilitation. However, in cases where treatment does not improve, the surgical option should be considered. Raising patient awareness and genetic consultation about prenatal diagnosis is important.

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Conflict of Interest: The authors declare no conflict of interest in this study.

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Author Contributions: N.Y.G. performed the physical examination of the patient, conducted a literature review and compiled it into an article.

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