Lumbar Spinal Stenosis Due to Ligamentum Flavum Hypertrophy in a Patient with Multiple Exostosis

Multipl Ekzostozu Olan Hastada Ligamentum Flavum Hipertrofisiyle Gelişen Lomber Spinal Stenoz

Spinal Stenosis in a Patient with Exostosis

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Özet

Herediter multipl ekzostoz genellikle genellikle uzun kemiklerin metafiz bölgelerini, sıklıkla da alt ekstremiteyi etkileyen birden fazla ekzostoz (osteokondrom) ile karakterize otozomal dominant bir hastalıktır ve nadiren aksiyel iskelette gelişir. Literatürde, spinal osteokondromlarla spinal kanal stenozunun geliştiği herediter multipl ekzostoz vakaları bildirilmiştir. Ancak herediter multipl ekzostoz olan bir hastada farklı bir hiperosteotik süreç olan ligamentum flavum hipertrofisi nedeniyle lomber spinal stenoz gelişebilir. Burada, böyle bir vakada patojenik mekanizmalar ve klinik özellikler ele alındı.

Anahtar Kelimeler

Multipl Herediter Ekzostoz; Spinal Stenoz; Ligamentum Flavum Hipertrofisi

Abstract

Hereditary multiple exostosis is an autosomal dominant disease characterized by multiple exostoses (osteochondromas) usually affecting the metaphysical regions of long bones, usually of the lower extremity, and seldom occurring in the axial skeleton. In the literature, hereditary multiple exostosis cases that developed spinal canal stenosis due to spinal osteochondromas have been reported. Lumbar spinal stenosis may occur in a hereditary multiple exostosis patient due to ligamentum flavum hypertrophy, which is a hyperosteotic process that differs from exostosis. We discuss one such case, along with pathogenetic mechanisms and clinical features.

Keywords

Multiple Hereditary Exostoses; Spinal Stenosis; Ligamentum Flavum Hypertrophy

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Introduction

The ligamentum flavum (LF) is a yellow elastic ligament that covers the spinal canal's posterior and lateral walls. It has several important roles, such as stabilizing the spine while sitting and standing, controlling intervertebral movements, and providing smoothness in the back wall of the spinal canal [1]. LF hypertrophy is an idiopathic or degenerative process closely associated with spinal stenosis [2].

Hereditary multiple exostosis (HME) is an autosomal dominant disease characterized by the presence of cartilaginous exostoses (osteochondromas) [3]. The basic defect is disturbed endochondral ossification with a large number of benign bone ossifications in juxtaepiphyseal areas where endochondral ossification is present. The incidence is 1/50,000 [4]. The most common locations are the distal femur and proximal tibia and, more rarely, the small bones of the hands and feet, pelvis, scapula, and vertebrae [5]. HME cases that developed spinal cord stenosis due to spinal exostoses have been reported in the literature [6,7]. However, as far as we know, development of spinal stenosis due to LF hypertrophy in a patient with HME has not been reported previously. We describe such a patient here.

Case Report

A 65-year-old female patient presented to our clinic complaining of pain in her low back and legs. Her symptoms especially appeared while standing and walking; she felt the need to bend forward while walking. She was a homemaker and had difficulty with household activities. She had gradually become unable to walk because of increased pain in both legs, and for those reasons she was admitted to our hospital. The claudication distance was around 15 metres. Additionally, the patient had limitation of motion in both knees and had often, from approximately the age of 5 or 6 years, experienced painless swellings at the inner surfaces of both knees. Her personal history revealed diabetes mellitus and hypertension. When the family history was queried, she reported similar swellings in the legs and elbows of her brother.

On physical examination, body weight was 65 kg and height was 142 cm. Her body mass index was calculated as 32 kg/m². The lumbar spine movements of the patient indicated that she had significant rotoscoliosis, limiting movement and mildly painful in all directions; this was more marked on extension. Her knee terminal extension and flexion were limited and the patellar mobility was decreased bilaterally. Valgum deformity was present in the right knee, mild varum deformity at the left knee, and calcaneovalgus deformity on the right. The left lower extremity was 3 cm shorter than the right. On neurological examination, hypoesthesia on the L4-L5 dermatome on the left was demonstrated. There was no motor weakness. Lower extremity deep tendon reflexes were hypoactive bilaterally but no pathologic reflex was found. Multiple, hard, immobile, and palpable masses were found in the lower extremities.

X-ray investigation revealed exostoses at the femur, distal fibula, and tibia (Fig.1). Lumbar vertebral magnetic resonance imaging (MRI) revealed findings of lumbar spondylosis and lumbar spinal stenosis at the L4-5 and L5-S1 levels due to hypertrophied LF and a large protrusion at the posterior L4-5 disc (Fig.2). Biochemistry, thyroid function tests, and vitamin B12 folic acid levels were within normal limits. The patient was thought to be suffering from HME and lumbar spinal stenosis and was placed into a physical treatment and rehabilitation program.

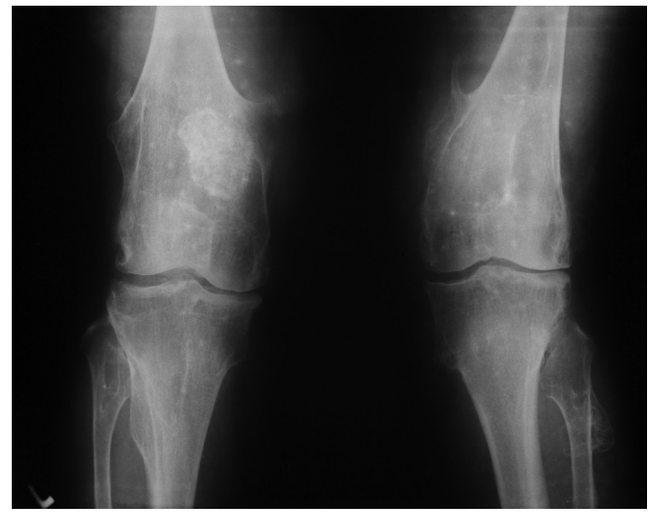


Figure 1. Plain radiograph of lower extremities showing exostoses.

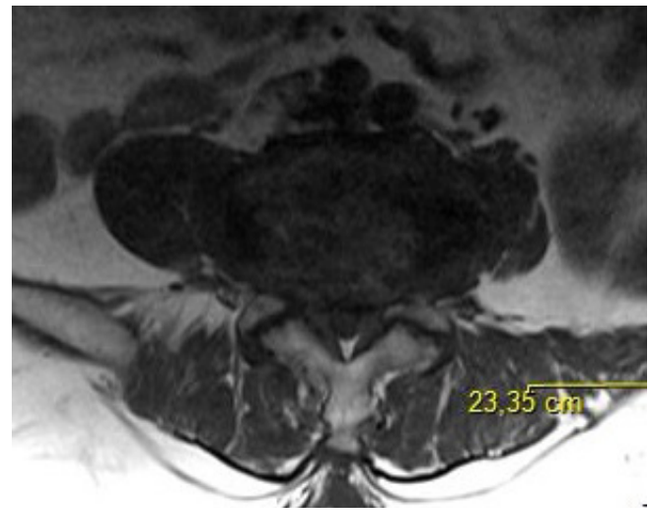


Figure 2. Magnetic resonance image showing degenerative changes in the faset joints, and stenosis of the spinal canal and foramen due to hypertrophy of the ligamentum flavum and a large protrusion at the posterior L4-5 disc.

Discussion

Lumbar spinal canal stenosis is a common cause of low back, lower extremity pain, and intermittent claudication, particularly in elderly patients. The LF covers an important part of the lateral and posterior walls of the spinal canal. Hypertrophy of LF results from LF degeneration brought about by tissue inflammation, fibrosis, and fibrocartilaginous transformation [8]. The histological changes in the hypertrophic LF have been reported as a loss of elastic fibers, increased collagen fibers, and chondrometaplasia, along with increased proteoglycans [9]. Degeneration and thickening of the LF can reduce the diameter of the spinal canal, compressing the dural sac and nerve roots, and contributing to low back pain [10].

HME is a disorder characterized by exocytosis that originates from the juxtaepiphyseal area of long bones. These exostoses, also called osteochondromas, originate from within the periosteum and grow progressively by endrochondral bone formation. Although the pathogenesis is still controversial, a disorder in

the EXT genes suppressing tumor growth and malignant transformation in the 8th, 11th, and 19th chromosomes has been suggested as playing a role [11]. To date, it is understood that EXT1 and EXT2 are tumor-suppressor genes and code a protein that participates in the biosynthesis of heparan sulfate. In patients with HME, heparan sulfate stays inside the cell and does not go through the membrane. This causes a disturbance in the negative feedback regulatory system, leading to the undifferentiated proliferation of chondrocytes in the metaphyseal region and resulting in the formation of osteochondromas [12].

Osteochondomas can be present in almost all bones except the skull bones. Although they are most commonly found in the metaphysis of long bones, 3-9% have been reported in the vertebrae [7]. HME is often clinically related to a skeletal deformity; the osteochondromas can cause pain, limitation in the range of motion, unequal extremity length, blood vessel and nerve suppression, valgus deformity in the knee and ankle joints, and rarely, osteosarcoma. HME often presents with neurological deficits because of direct compression of the peripheral nerve, nerve root, or rarely, the spinal cord [12].

HME is diagnosed by radiologically seeing a minimum of 2 exostoses in the juxtepiphyseal areas of long bones together with a positive family history and/or demonstration of a gene mutation [3]. On plain radiographs, in a long bone, an osteochondroma typically appears as a pedunculated or sessile bone-like projection. According to this guideline, the patient was thought to be suffering from HME. The clinical symptoms indicated lumbar spinal stenosis and the patient was diagnosed with HME, as a vertebral osteochondroma can also cause vertebral stenosis. It is difficult to make a diagnosis with plain radiography due to the complex image created in the spine by spinal osteochondomatosis.

Computed tomography (CT) scan is one of the diagnostic imaging modalities. It demonstrates the extent of the cartilaginous and osseous components and their relationship with the vertebral and neural elements of the spine. But MRI also allows direct visualization of the cord and nerve roots and thus is more useful than CT in defining the extradural component and cord compression [13]. The lumbar vertebral MRI of our patient revealed stenosis at the spinal canal together with more significant LF hypertrophy at the L4-5 disc level.

In conclusion, our patient who had a symptom of pain in the low back and legs and who described neurogenic claudication was diagnosed with lumbar spinal stenosis and HME. Spinal stenosis was seen to have developed due to the LF hypertrophy, a hyperosteotic process that differs from HME. Although conventional radiography is enough to confirm the HME diagnosis and to determine the level of disease, an MRI should be the imaging method of choice to evaluate symptomatic lesions and complications.

Competing interests

The authors declare that they have no competing interests.

References

1. Kasım FB, Tosun O, Kasım E, Ercan K, Tosun A, Arslan H, et al. "Thickened" ligamentum flavum caused by laminectomy. Neurol Neurochir Pol 2015;49(3):145-9.
2. Altinkaya N, Yildirim T, Demir S, Alkan O, Sarica FB. Factors associated with the thickness of the ligamentum flavum: is ligamentum flavum thickening due to hypertrophy or buckling? Spine (Phila Pa 1976). 2011;36(16):E1093-7.
3. Pannier S, Legeai-Mallet L. Hereditary multiple exostoses and enchondromatosis. Best Pract Res Clin Rheumatol 2008;22(1):45-54.
4. Akbal A, Kurtaran A, Selçuk B, Kimyon S, Akyüz M. A Case with Multiple Hereditary Exostoses. J PMR Sci 2010;13:34-6.
5. Aldea S, Bonneville F, Poirier J, Chiras J, George B, Carpentier A. Acute spinal cord compression in hereditary multiple exostoses. Acta Neurochir (Wien) 2006;148(2):195-8.
6. Giudicissi-Filho M, de Holanda CV, Borba LA, Rassi-Neto A, Ribeiro CA, de Oliveira JG. Cervical spinal cord compression due to an osteochondroma in hereditary multiple exostosis: case report and review of the literature. Surg Neurol 2006;66(Suppl.3):S7-11.
7. Bess RS, Robbin MR, Bohlman HH, Thompson GH. Spinal exostoses: analysis of twelve cases and review of the literature. Spine (Phila Pa 1976) 2005;30(7):774-80.
8. Xu YQ, Zhang ZH, Zheng YF, Feng SQ. MicroRNA-221 Regulates Hypertrophy of Ligamentum Flavum in Lumbar Spinal Stenosis by Targeting TIMP-2. Spine (Phila Pa 1976) 2016;41(4):275-82.
9. Kosaka H, Sairyo K, Biyani A, Leaman D, Yeasting R, Higashino K, et al. Pathomechanism of loss of elasticity and hypertrophy of lumbar ligamentum flavum in elderly patients with lumbar spinal canal stenosis. Spine (Phila Pa 1976) 2007;32(25):2805-11.
10. Sairyo K, Biyani A, Goel V, Leaman D, Booth R Jr, Thomas J, et al. Pathomechanism of ligamentum flavum hypertrophy: a multidisciplinary investigation based on clinical, biomechanical, histologic, and biologic assessments. Spine (Phila Pa 1976) 2005;30(23):2649-56.
11. Stieber JR, Dormans JP. Manifestations of hereditary multiple exostoses. J Am Acad Orthop Surg 2005;13(2):110-20.
12. Guo XL, Deng Y, Liu HG. Clinical characteristics of hereditary multiple exostoses: a retrospective study of mainland chinese cases in recent 23 years. J Huazhong Univ Sci Technolog Med Sci 2014;34(1):42-50.
13. Chooi YS, Siow YS, Chong CS. Cervical myelopathy caused by an exostosis of the posterior arch of C1. J Bone Joint Surg Br 2005;87(2):257-9.

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