Spinal Stenosis in a Patient with Exostosis

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

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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 hyperostotic 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
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 spondylolisthesis 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 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.

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 periosseous and grow progressively by endochondral 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].

Osteochondromas 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 of movement, 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 juxtephyseal 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 osteochondromatosis.

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

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