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Case Report

A case of myelopathy because of enchondromas from Maffucci syndrome with successful surgical treatment

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Abstract

BACKGROUND CONTEXT: Maffucci syndrome is a rare disorder comprising multiple enchondromas associated with multiple hemangiomas. Less than 200 cases have been reported in the literature. Most reported cases comprised lesions of the long bone metaphyses, hands, and feet. No previous case of myelopathy due to enchondroma from Maffucci syndrome has been reported.

PURPOSE: To highlight an interesting and rare presentation of thoracic myelopathy because of enchondromas from Maffucci syndrome.

STUDY DESIGN: This is a case report of a single patient presenting with myelopathy in whom enchondromas from Maffucci syndrome were removed from the spinal canal with the return of normal function.

METHODS: Clinical examination, magnetic resonance imaging, surgical resection, and histologic analysis.

RESULTS: The patient experiencing myelopathy symptoms underwent a surgical resection of the offending lesions that resulted in return to normal function.

CONCLUSIONS: Maffucci syndrome is a rare condition; however, in affected patients the possibility of structural spinal abnormalities causing cord compression must be considered, as discrete surgical resection can result in a good outcome.

Keywords: Myelopathy; Maffucci syndrome; Enchondroma; Spinal cord; Thoracic; Tumor

Introduction

Maffucci syndrome is an extremely rare nonhereditary condition comprising multiple hemangiomas and enchondromas. The few cases reported in the English language literature comprised primary lesions of the metaphyseal long bones, hands, and feet. We present a case of enchondromas from Maffucci syndrome compressing the spinal cord and presenting as myelopathy.

Case presentation

A 39-year-old man presented complaining of balance and coordination difficulty. He had been previously active and played softball competitively until 2 years before the presentation. He initially noted a ‘‘freezing’’ and ‘‘tingling’’ sensation in his feet bilaterally that was difficult to localize. Over the next several months, he began having difficulty in maintaining balance while running. This progressed to include balance difficulty while walking. He reported being able to walk up and down stairs normally 7 months before presentation, but now was unable to do so without assistance. At the time of presentation, the patient was noted to walk with an ataxic wide-based gait. He complained of difficulty in performing fine motor tasks, and his deficits had progressed to the point that he was having great difficulty with both normal ambulation and performing activities of daily living.

The patient’s medical history was significant only for Maffucci syndrome, with which he had been diagnosed at 17 months. He had multiple enchondromas and had undergone multiple procedures of his legs for leg length discrepancy. Gross deformity typical of Maffucci syndrome, including multiple hard nontender nodules, were present on the patient’s hands and feet bilaterally. These had

FDA device/drug status: Not applicable.

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minimal functional impact on the patient's normal activities or athletic participation.

On physical examination, the previously mentioned visible nontender swellings of the bilateral metacarpals and metatarsals were noted. Motor strength was assessed and noted to be Grade 5 in all muscle groups of the upper extremities bilaterally. All groups in the lower extremities were also Grade 5, with the exception of the right iliopsoas, which was graded 4+/5. The patient was hyperreflexic (3+) to bilateral patellar and Achilles reflex testing. Upper extremity reflexes were 1+. No Hoffman sign was noted, but Babinski sign was present bilaterally. In addition, sustained clonus was elicited in both lower extremities. The previously mentioned ataxic gait was again noted. Based on these findings, the patient was felt to be Grade 3 on the Nurick classification for cervical myelopathy [1]. This was based on the patient's ability to walk without assistance along a straight floor, but inability to navigate the stairs, which affected his ability to work full time.

The patient underwent X-ray, computed tomography, and magnetic resonance imaging of the cervical, thoracic, and lumbar spine. Multiple congenital fusion anomalies were noted throughout the posterior elements of the cervical spine. At the cervical C5–C6 level, there was a large disc osteophyte complex with advanced degenerative changes. The multiple spontaneously fused adjacent segments likely contributed to the advanced degeneration noted (Figs. 1 and 2). There was cord compression on the left side with increased signal on the sagittal T2 images (Fig. 3). In the thoracic spine, multiple levels beginning at T2 and extending to T6 were noted to have fused posterior elements, including both laminae and spinous processes (Fig 4). A large exostosis arose from the vertebral bodies and left facet joint at T5/T6 and extended medially into the spinal canal (Fig. 5). This resulted in severe compression of the spinal cord, displacing the cord from left to right into the neuroforamen. Marked edema and myelomalacia within the cord at the T5–T6 level was noted (Figs. 6 and 7).

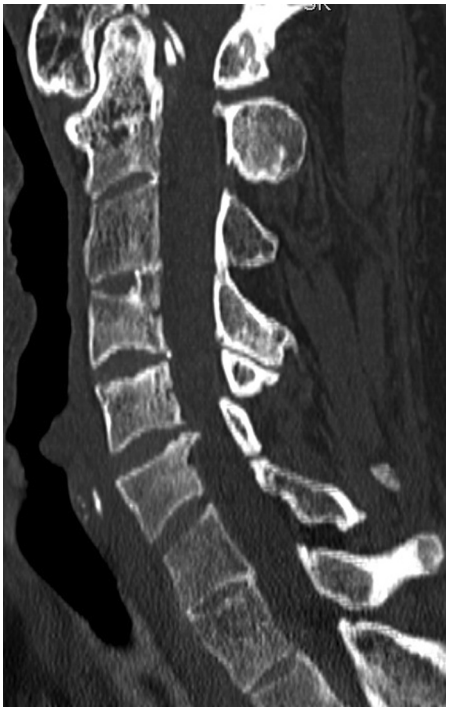

Fig. 1. Sagittal computed tomography of the cervical spine showing a disc/osteophyte complex at C5/C6.

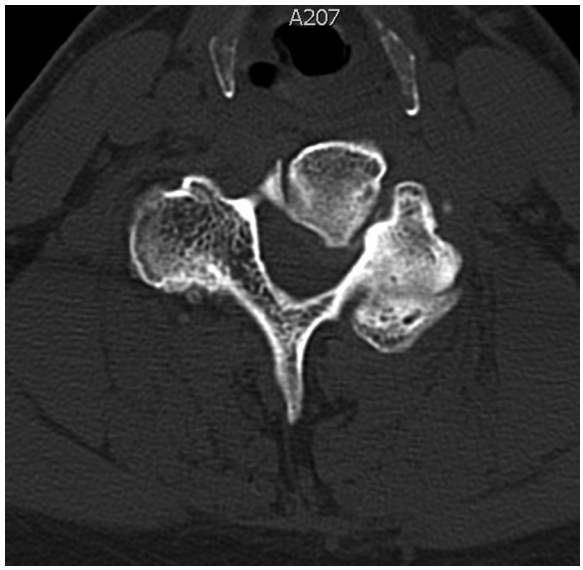

Fig. 2. Axial computed tomography of the cervical spine showing a disc/osteophyte complex at C5/C6.

Given the patient's concomitant cervical and thoracic involvement, the decision to stage surgical intervention was made. Because of the anticipated easier recovery, the cervical spine was addressed first with an anterior cervical decompression and fusion with partial corpectomies at C5–C6. This procedure and its recovery were uneventful. By postoperative Week 6, the patient noted that his ambulation felt steadier, but significant ataxia was still present.

On postoperative Week 8 after the cervical decompression and fusion, the thoracic spine lesion was addressed. Given the location and morphology of the mass, both posterior and far lateral approaches were used to perform a costotransversectomy, transpedicular partial vertebrectomy, and mass excision. In the operating room, the lamina showed multiple levels of congenital abnormalities as were visualized on the preoperative studies. Pedicle screws were placed at the T2, T3, T7, and T8. A wide multilevel laminectomy was then undertaken from T2 to T8 to allow for

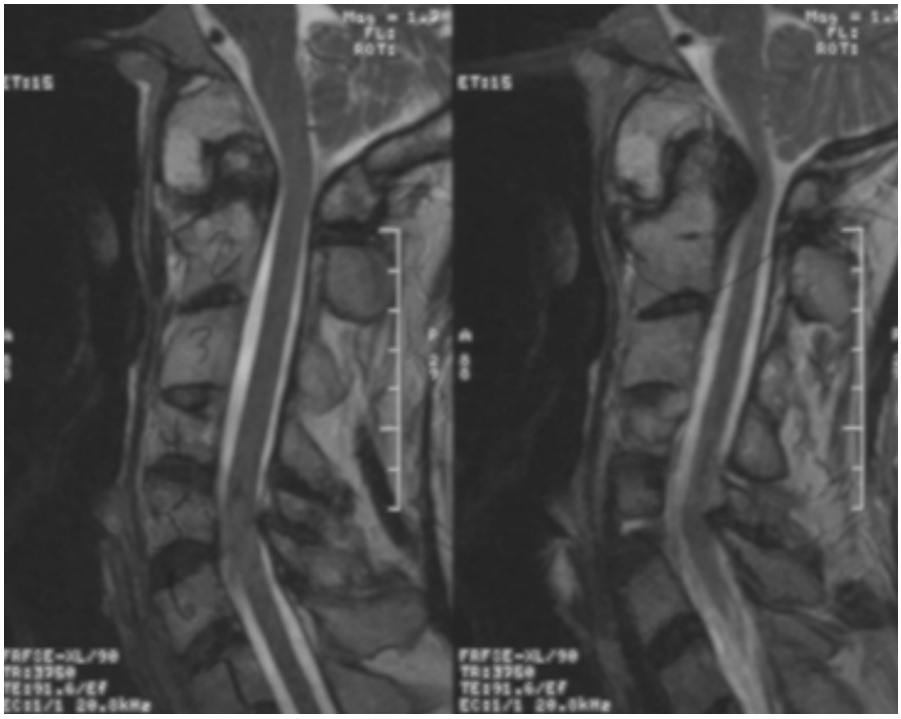

Fig. 3. Sagittal T2-weighted magnetic resonance imaging sequence showing cord compression at the C5/C6 level.

ample space for the cord during the transpedicular decompression. At the T5/T6 level, transpedicular exposure was used to visualize the pedicles, which were removed in a medial to lateral fashion. The large enchondroma noted on imaging was visualized on the left at the T5/T6 level compressing the cord. The T5 nerve root was also severely compressed. A costotransversectomy and extracavitary far lateral exposure was performed, followed by decompression and partial vertebrectomy at the T5/T6 level. This was continued down to the disc space verifying that the posterior aspect of the vertebral body was identified. The mass was freed off the dura, excised, and sent for pathologic evaluation. Postoperative imaging is provided (Fig. 8).

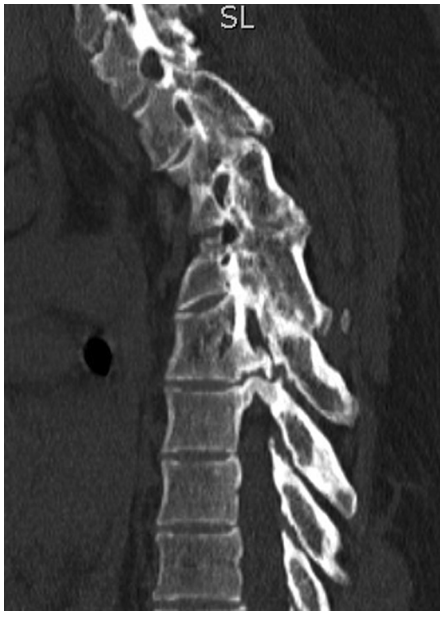

Fig. 4. Sagittal computed tomography of the thoracic spine showing congenital fusion of the posterior elements at multiple levels.

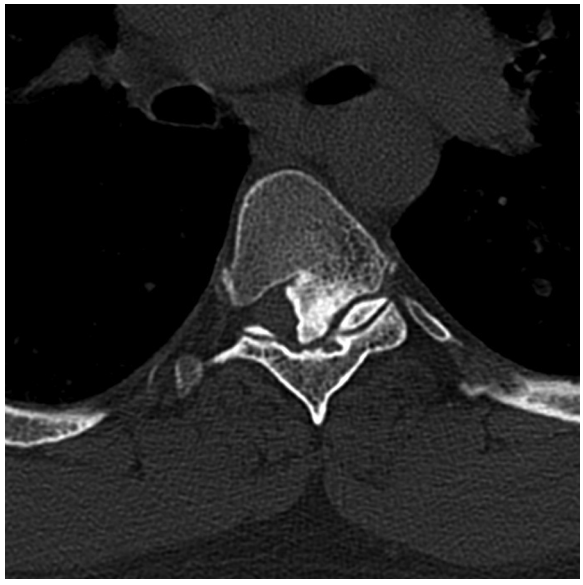

Fig. 5. Axial computed tomography of thoracic spine showing a large exostosis encroaching the spinal canal at the T5/T6 level.

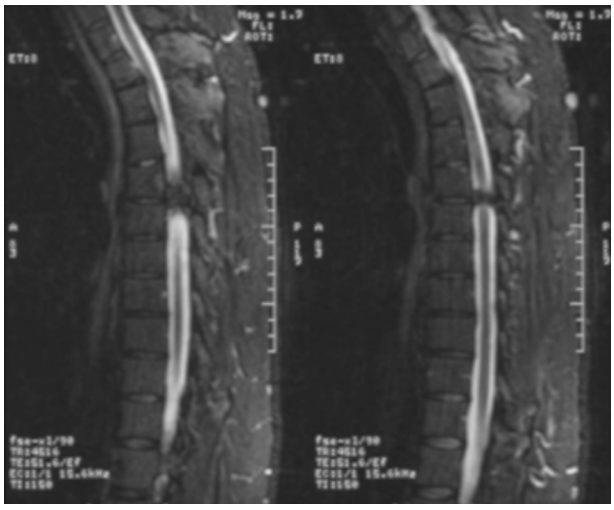

Fig. 6. Sagittal T2 magnetic resonance imaging showing cord compression at the T5/T6 level.

Pathology of the epidural mass showed fragments of lamellar bone and reactive hyaline cartilage consistent with an enchondroma. The bone taken for the laminectomy was also sent to pathology and showed fragments of intervertebral disc and reactive hyaline cartilage.

Immediately postoperatively, the patient's neurologic status was stable. He had an uncomplicated postoperative course and was discharged without incident. At 8 months postoperative follow-up, his function was significantly improved. His upper extremity fine motor function had returned to normal. He was able to ambulate without assistance, although some mild ataxia was still present. At this time we classified his Nuric grade as 2.

Discussion

Maffucci syndrome is a congenital nonhereditary disorder comprising a combination of multiple enchondromas with multiple hemangiomas. It is an extremely rare disorder, with less than 200 cases reported in the English language literature [2]. The syndrome was first described in an autopsy report by Maffucci in 1881, when a 40-year-old woman died after amputation of an arm for multiple bleeding vascular tumors. A more complete description with naming of the syndrome for Maffucci was provided by Carleton in 1942 [3].

Children with Maffucci syndrome are typically normal at birth, but before puberty are thought to undergo mesodermal dysplasia that leads to both skeletal and vascular malformations [4]. The skeletal lesions are most commonly enchondromas with a predilection for the metaphyseal regions of the long bones and phalanges; however, involvement of the ribs, vertebrae, and skull has been reported [5,6]. Significant growth disturbance and bony deformity are common, with the hands and feet being the areas most affected. The enchondromas present in Maffucci syndrome carry a significant risk of malignant transformation, most commonly to chondrosarcoma. Several case series have shown the risk of malignancy to range from 23% to 59% [7].

The vascular component of Maffucci usually consists of hemangiomas, often of the cavernous type that form calcified thrombi known as phleboliths. Lymphatic malformations have been reported as well [8]. The hemangiomas typically present as blue, grape-like swellings on the distal upper and lower extremities that can be emptied by pressure. They have been known to occur in the colon and brain [9]. Sarcomatous degeneration of these lesions has been reported, associated with central nervous system, pancreatic, and ovarian malignancies [10].

To our knowledge, there have been no previous reports in the literature of skeletal lesions from Maffucci syndrome presenting with cervical or thoracic myelopathy. Patients with this rare disorder must be closely monitored not only for malignant transformation, but also for morbidity from the local effects of their benign skeletal and vascular lesions, and structural spinal lesions must

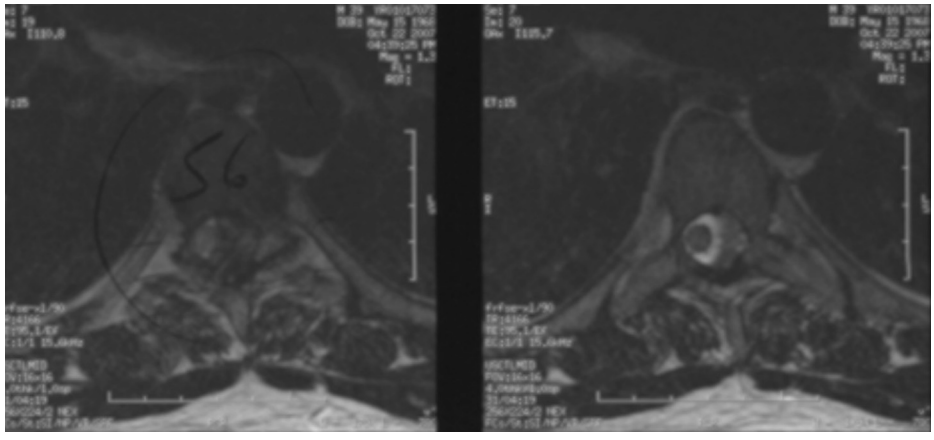

Fig. 7. Axial T2 magnetic resonance imaging showing cord compression at the T5/T6 level.

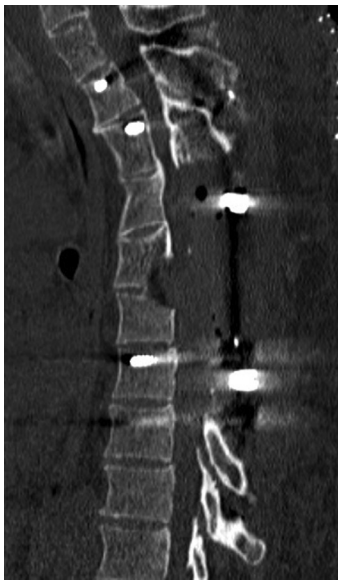

Fig. 8. Postoperative sagittal computed tomography of the thoracic spine showing laminectomy, excision of mass, and partial vertebrectomy at the T5/T6 level with instrumented fusion.

always be considered when these patients present with neurologic complaints.

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