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Case Report

Multiple hereditary osteochondroma presenting as spastic paraplegia

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ABSTRACT

Osteochondromas are benign bony tumours with only 1–4% being located in the spine. Compressive myelopathy as a presentation of vertebral osteochondroma in a child is a rare entity. Our case being 6-year-old female with multiple osteochondroma involving spine presenting as spastic paraplegia is itself rare as males of 2nd decade are mostly affected. Father of the patient also had osteochondroma like lesion over knee showing its autosomal dominant inheritance. Appropriate imaging followed by surgical decompression is key to minimize further complications.

Keywords- Osteochondroma, Compressive myelopathy, Spastic paraplegia, Multiple exostosis

INTRODUCTION

Hereditary multiple exostoses also called hereditary multiple osteochondromas, is a rare genetic disorder characterized by multiple osteochondromas that grow near the growth plates of bones such as ribs, pelvis, vertebrae and especially long bones. Children commonly present at 5-15 years of age. It is autosomal dominant disorder due to pathogenic variants in EXT1 or EXT2, characteristically benign, with only 1-5% progressing to chondrosarcoma. They appear as bony protrusions with a cartilaginous cap, most typically along the metaphysis of long bones. Solitary lesions are the most common in individuals diagnosed with osteochondroma.[1]

This is a cartilage capped exophytic lesion that grows on external surface of the bone containing a marrow cavity continuous with that of parent bone. Osteochondroma stops growing at the physeal maturation age (unless malignant change has occurred). Osteochondromas have also been shown to regress.[2] Neurological symptoms due to myelopathy or radiculopathy are seldom present as most lesions grow outside the spinal canal (extramedullary lesion).[3]

CASE REPORT

A 6-year-old female child, product of a non-consanguineous marriage, 3rd in order presented with complaints of inability to walk since last 6 months. Weakness of bilateral lower limb insidious in onset, initially started from right lower limb followed by left, slowly progressing and resulted in complete inability to walk and sit over a period of 6 months. No history of previous similar complaints, facial weakness, vision disturbances, altered sensorium, fever, headache, vomiting, loss of consciousness and seizures.

No history of bladder and bowel disturbance was present. No history of any trauma or recent viral infections or TB contact in family. Child had two swellings, one in the midline on back of neck and another on left side of neck since birth and slowly progressing in size since then. There was significant family history of similar swelling on left knee of father of approximately 3×3 cm size. The developmental milestones including motor milestone were achieved at normal age and there was no history of regression of milestones. Patient was completely immunized including polio vaccination.

On examination

General physical examination revealed normal vitals, normal built and weight, height and BMI were appropriate for age. It didn't reveal any abnormality except bony swellings over neck and knee.

CNS examination

Higher mental functions were intact. No cranial nerve deficits. Motor system examination showed bulk of Left lower limb more than right, spasticity of bilateral lower limbs with power 0/5 in bilateral lower limbs and trunk muscles, power 5/5 in upper limb girdle. Knee jerk and ankle jerk were exaggerated and plantar bilateral extensor. Biceps and triceps jerk were normal. No sensory disturbances, no signs of meningeal irritation, no bladder and bowel disturbance were present.

Skull and spine examination revealed a hard bony, non-tender, non-fluctuant swelling of 3×3 cm at the cervical region in the midline and a similar swelling on the left lateral aspect of neck of size 2×2 cm (Figure 1). On detailed examination one more bony swelling of 1×1 cm was found on the lateral aspect of right knee joint. All the routine investigations were done and all parameters were within normal range including ALP. Sepsis screen was negative. X-ray and USG spine done which shows bony swelling at the cervical spine region of size 6×4×4 cm. X-ray Knee shown metaphyseal exostosis of right femur growing away from growth plate (Figure 2).

MRI spine was done to diagnose any spinal deformity which shown large lesion of size 67CC×46AP×43LT mm arising from left lamina, left transverse process and spinous process of C5, C6, C7, D1, D2 and D3 vertebrae and seen extending into spinal canal at D1-D2 level and pushing and compressing spinal cord anteriorly S/o Osteochondroma (Figure 3). Child was referred to Neurosurgery department for surgical intervention.

Figure 1: Swellings over midline and left lateral side of neck.

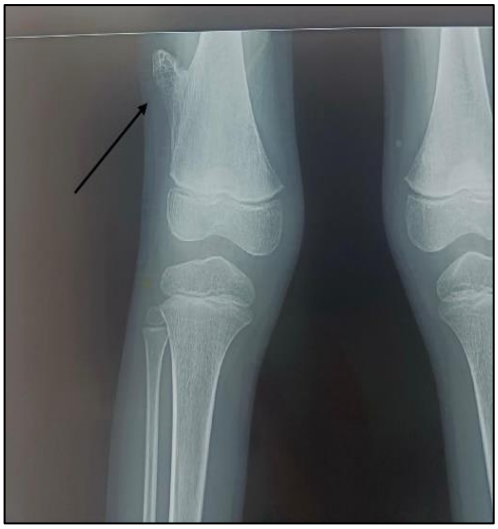

Figure 2: X-ray right knee joint showing metaphyseal exostosis of femur.

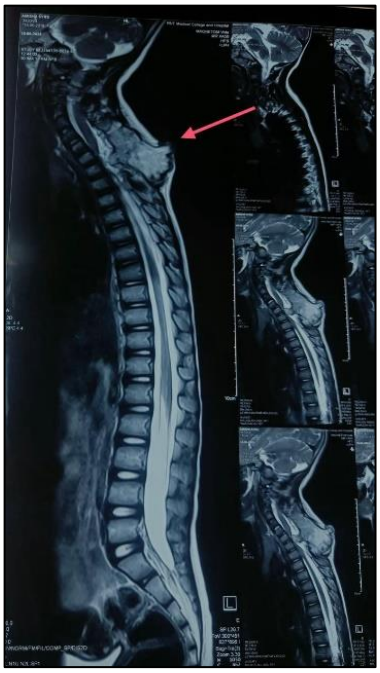

Figure 3: MRI spine showing bony overgrowth in cervical and thoracic spine compressing spinal cord.

DISCUSSION

Osteochondroma is one of the common benign tumors usually found in the appendicular skeleton. Between 1.3% and 4.1% of solitary osteochondroma arise in the spine. Osteochondroma of the spine usually arises from the neural arch of the cervical and thoracic vertebra and can cause compression of the spinal cord.[4]

The incidence of osteochondroma is 1 in 50,000. The long bones are most affected; only 1–4% of osteochondromas are found in the spinal column. They are more common in men with a ratio of 1.5:1.[5]

Osteochondroma is rare in children and according to many studies conducted between 1969 to 2020 in children up to 18 years of age, only 2 studies had children <7 years presented with osteochondroma involving spinal cord.[6] Previously reported case of 6-year-old girl with features of progressive gait imbalance, lower limb weakness and bladder / bowel disturbance for 3 months. She had spastic paraparesis with motor and sensory levels of D2-3 and with sphincter involvement along with multiple bony hard swellings. CT and MRI revealed osteochondroma arising from C7, D1, D2 pedicles, compressing the thecal sac.[7] Another case of a 9-year-old boy with an acute history of frequent falls while walking and standing. Hypertonia in both lower limbs and spasticity was positive. Local examination did not reveal any visible swelling or deformity except for tenderness at D1-D2 spinous processes. Plain CT of thorax revealed anterior wedge compression of D1 vertebral body. MRI of the whole spine revealed an enhancing extradural mass protruding into the spinal canal at T1 vertebral level causing compression, and posterior displacement of spinal cord.[4]

Our case is rare as osteochondroma was found in a female child of age less than 7 years. Differential diagnosis includes pott's spine, osteosarcoma, extradural abscess. In general, cord compression tends to be rare as most lesions grow out of the spinal canal. The reported incidence of myelopathy or radiculopathy caused by the tumour is 0.5–1%.[3]

CONCLUSION

In children with spinal cord compression due to what appears to be a bony lesion on imaging, osteochondroma should be considered in the differential diagnosis as these lesions are not as rare as once thought. Appropriate imaging followed by surgical decompression is key to minimizing the risk of permanent neurologic deficits, tumor recurrence, and, although rare, malignant transformation.

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