CASE REPORT

Unusual case of hereditary multiple exostoses presenting with compressive myelopathy

Kumar Amerendra Singh,¹ Vijay Shankar,¹ Simanchal P Mohanty²

¹Department of Orthopaedics, Kasturba Medical College Manipal, Manipal, Karnataka, India

²Department of Orthopaedics, Manipal University, Manipal, Karnataka, India

Correspondence to
Dr Kumar Amerendra Singh,
amerendra2@gmail.com

Accepted 20 April 2016

SUMMARY

A 15-year-old boy presented with progressive weakness of both lower limbs. He had classical clinical features suggestive of multiple hereditary exostoses (MHE). Imaging studies of the spine revealed osteochondromas at D7, D11 and L5 vertebrae. The tumour at D11 was encroaching into the spinal canal and causing cord compression. The patient underwent osteochondroma excision and posterior spinal stabilisation, following which there was complete neurological recovery by 3 months.

BACKGROUND

Multiple hereditary exostoses (MHE) frequently involve the appendicular skeleton, such as at the metaphyseal area of the long bones, iliac crest and scapula. Involvement of the spine is uncommon, with an incidence of 1–9%.^{1 2} Though there are reports of solitary osteochondroma causing spinal cord compression, involvement of the cord in patients with MHE is rare.³ MHE involving the spine at different levels is also not common.^{3 4} We report an unusual case of MHE with osteochondromas at D7, D11 and L5 vertebral levels presenting with compressive myelopathy.

CASE PRESENTATION

A 15-year-old boy presented with difficulty in walking and progressive weakness in both lower limbs of 3-month duration. The patient's father and brother also had features of MHE. The boy had clinical features of multiple exostoses with deformities of the wrist joints, knee joints and ankle joints. On examination of the back, three hard bony swellings were present in the paraspinal area. Neurologically, the boy had grade 3 power (Medical Research Council) in both lower limbs, without sensory deficit.⁵ Knee and ankle jerks were exaggerated with plantar reflex showing a Babinski response. There was neither bowel nor bladder involvement.

INVESTIGATIONS

Plain radiographs showed features suggestive of diaphyseal aclasia (figure 1). CT scans and MRI of the spine showed bony masses arising from the posterior elements of D7, D11 and L5 vertebrae. The mass arising from the right pedicle of D11 vertebra was encroaching into the spinal canal, compressing the cord (figure 2), whereas tumours involving the left lamina of D7 and spinous process of L5 were not extending into the spinal canal (figure 3).

Figure 1 Radiographs showing multiple exostoses involving proximal and distal ends of both femora and bilateral proximal tibia.

TREATMENT

The 4×3×3 cm hard bony mass, arising from the right pedicle of the D11 vertebra, encroaching into the spinal canal and extending anteriorly, laterally and posteriorly, was excised (figure 4). Following excision of the tumour, posterior decompression of the cord was carried out. Intraoperatively, spinal cord pulsation was present. The mass arising from the left lamina of D7 vertebra was also excised. This was supplemented with posterior spinal instrumentation.

OUTCOME AND FOLLOW-UP

Histopathological examination confirmed the diagnosis of benign osteochondromas. The patient recovered gradually and at the end of 3 months had grade 5 power in both lower limbs. At 2 years follow-up, he had normal locomotor function and was able to carry out all activities of daily living (figure 5).

DISCUSSION

Boyer first described MHE in 1814. Mutation of EXT genes that code for a protein involved in biosynthesis of heparan sulfate leads on to

Figure 2 T2-weighted thoracic spine MRI showing osteochondroma arising from right pedicle of D11 vertebra compressing the spinal cord.

To cite: Singh KA, Shankar V, Mohanty SP. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2016-215440

Figure 3 CT scan of the spine showing osteochondromas arising from D7 (A), D11 (B) and L5 (C) vertebrae.

undifferentiated proliferation of chondrocytes in the metaphyseal region.⁶ Vertebral lesions have a predilection for the cervical spine (50–80%) followed by the thoracic (20–36%) and, rarely, the lumbar region.³ Extracanal lesions usually present as palpable masses, while intracanal lesions may present with gait disturbances due to compressive myelopathy.⁷ This patient presented with multiple painful swellings over the back and progressive weakness of the lower limbs indicating cord compression.

Incidence of MHE involving multiple vertebral levels is uncommon with few cases reported.^{3–4} Compressive myelopathy is more commonly seen with solitary osteochondroma than in MHE.^{1–7} In our case, the tumour at the D11 vertebra was the cause of spinal cord compression and hence excised. The osteochondromas at D7 and L5 were extraspinal. The tumour at D7 was excised because it was relatively large and painful. Since the

Figure 4 Intraoperative photograph showing osteochondroma being excised from D11 pedicle.

Figure 5 Follow-up radiographs and CT scan at 2 years showing no recurrence.

right pedicle along with the lamina was removed; posterior spinal instrumentation was performed to prevent the development of deformity.

The treatment of choice for symptomatic spinal osteochondromas is surgical excision.^{1–4} When presenting with features of compressive myelopathy, excision of the tumour results in good recovery of neurological function with less chance of recurrence. This patient recovered completely, without residual neurological deficit.

All patients with MHE should be screened for spinal lesions. They should be followed up at regular intervals to detect early features of spinal cord compression.

Learning points

- ▶ This is a rare case of multiple hereditary exostoses (MHE) with spinal osteochondromas occurring at multiple vertebral levels and producing compressive myelopathy.
- ▶ Removal of spinal osteochondromas causing cord compression produces excellent neurological recovery.
- ▶ Removal of tumours from posterior elements and pedicles should preferably be supplemented with posterior spinal stabilisation to prevent progressive deformity.
- ▶ Even though MHE involving the spine are rare, all patients should be screened to rule out lesions in the spine so that compressive myelopathy can be prevented.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Quirini GE, Meyer JR, Herman M, et al. Osteochondroma of the thoracic spine: an unusual cause of spinal cord compression. *AJNR Am J Neuroradiol* 1996;17:961–4.
- 2 Roach JW, Klatt JW, Faulkner ND. Involvement of the spine in patients with multiple hereditary exostoses. *J Bone Joint Surg Am* 2009;91:1942–8.
- 3 Ashraf A, Larson AN, Ferski G, et al. Spinal stenosis frequent in children with multiple hereditary exostosis. *J Child Orthop* 2013;7:183–94.

- 4 Potocki K, Prutki M, Kralik M, et al. Spinal cord compression caused by multiple spinal osteochondromas. *Wien Klin Wochenschr* 2008;120:538.
- 5 Medical Research Council. *Aids to the examination of the peripheral nervous system*. Memorandum no. 45. London: Her Majesty's Stationery Office, 1981.
- 6 Cannon JF. Hereditary multiple exostosis. *Am J Hum Genet* 1954;6:419–25.
- 7 Palmer FJ, Blum PW. Osteochondroma with spinal cord compression: report of three cases. *J Neurosurg* 1980;52:842–5.

Copyright 2016 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow