Clinical Neurology and Neurosurgery 118 (2014) 5–8

Contents lists available at ScienceDirect

Clinical Neurology and Neurosurgery

journal homepage: www.elsevier.com/locate/clineuro

Case report

Multiple spinal osteochondromata and osteosarcoma in a patient with Gorlin's syndrome

Ahmed-Ramadan Sadek[a,c,*,1,2], Girish Vajramani[a,c,1], Simon Barker[a,c], Mark Walker[a,c], Colin Kennedy[b,c], Ali Nader-Sepahi[a,c]

[a] Department of Neurosurgery, Wessex Neurological Centre, University Hospital Southampton NHS Foundation Trust, Tremona Road, Southampton SO16 6YD, UK
[b] Department of Pediatric Neurology, Wessex Neurological Centre, University Hospital Southampton NHS Foundation Trust, Tremona Road, Southampton SO16 6YD, UK
[c] Division of Clinical Neurosciences, School of Medicine, University of Southampton, Tremona Road, Southampton SO16 6YD, UK

ARTICLE INFO

Article history:
Received 22 August 2013
Received in revised form 23 October 2013
Accepted 29 November 2013
Available online 12 December 2013

Keywords:
Osteochondroma
Osteosarcoma
Gorlin's syndrome
Spine

1. Introduction

Gorlin's syndrome also known as nevoid basal cell carcinoma syndrome (NBCCS) is characterised by multiple basal cell carcinomas, benign jaw keratocystic odontogenic tumours, palmar/plantar pits, calcification of falx cerebri, and spine and rib anomalies [6]. This constellation of pathologies has been noted as far back as Ancient Egypt and its current prevalence in the United Kingdom is approximately 1 per 55,000 [3]. The syndrome results from mutations in the PTCH1 gene which is located on the long arm of chromosome 9 and is transmitted in an autosomal dominant manner [3]. Although various spinal anomalies such as calcification of nuchal ligament, fusion of vertebrae, hemivertebra and kyphoscoliosis have been described in this condition, the occurrence of multiple spinal osteochondromata and or osteosarcoma has not been reported [6,7]. We present a female patient with Gorlin's syndrome who developed thoracic and lumbar osteochondromata and recurrent sacral osteosarcomas.

2. Case report

At the age of 4 years the female patient underwent resection of vermian medulloblastoma, which was followed by craniospinal irradiation. Three years later she developed multiple BCCs of the scalp and was diagnosed with Gorlin's syndrome. She underwent numerous resections of the scalp BCCs. At the age of eight she presented with a palpable painless lump over her lower back and was diagnosed to have a L2 spinous process osteochondroma on computed tomographic (CT) scan (Fig. 1a). This was initially managed conservatively with serial follow-up scans. At the age of 13 years the lesion was excised after it was noted to have enlarged significantly (Fig. 1b). The diagnosis of osteochondroma was confirmed on histological examination (Fig. 2a). Radiographic imaging at the time also demonstrated an incidental intraspinal T9 lesion that was assumed to be a further osteochondroma but in the absence of symptoms this was managed conservatively. Three years later she re-presented with left lower limb stiffness and examination revealed increased tone, hyperreflexia, an extensor plantar response with normal power and sensation. The

* Corresponding author at: Department of Neurosurgery, Wessex Neurological Centre, University Hospital Southampton NHS Foundation Trust, Tremona Road, Southampton SO16 6YD, UK. Tel.: +44 23 80794084.
E-mail address: a.sadek@soton.ac.uk (A.-R. Sadek).
[1] These authors contributed equally to the manuscript.
[2] The Corresponding author has the right to grant on behalf of all authors and does grant on behalf of all authors, an exclusive licence (or non exclusive for government employees) on a worldwide basis to the Elsevier Publishing Group Ltd., to permit this article to be published in CNN and any other CNN products and sublicences such use and exploit all subsidiary rights, as set out in our licence.

http://dx.doi.org/10.1016/j.clineuro.2013.11.022

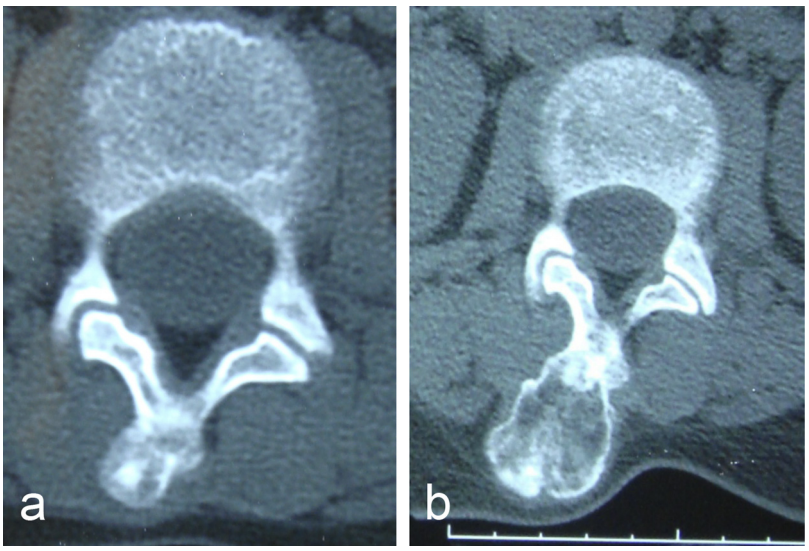

Fig. 1. (a) Axial CT of the lumbar spine shows the L2 spinous process osteochondroma. (b) Follow up CT showing considerable enlargement of the tumour.

contralateral lower limb had no neurological deficit. Magnetic resonance imaging (MRI) and CT scan demonstrated an intraspinal left sided lesion at T9 arising at the junction of the vertebral body and pedicle with compression of the spinal cord (Fig. 3a–c). A hemilaminectomy was performed and the tumour was resected via the transpedicular approach. Histological examination confirmed an osteochondroma. Postoperatively her stiffness and abnormal neurological signs resolved. Follow-up MRI scan confirmed adequate resection and absence of cord compression. During the same year she developed further basal cell carcinomas, an ovarian cyst, epilepsy, intracranial falx calcifications and multiple jaw cysts – all manifestations of Gorlin's syndrome. Four years later she developed back pain radiating to left buttock and leg in the absence of neurological deficit. A MRI scan revealed an intraspinal mass on the left side between S1 and S2 encasing the S2 nerve root (Fig. 3d). She underwent S1-S2 Laminectomy and excision of the lesion. Histopathology revealed it to be an osteosarcoma (Fig. 2b). Over the following twelve months she developed recurrence of the sacral osteosarcomas and underwent a sacrectomy at the National Orthopaedic Hospital. During the post-operative period she developed acute respiratory distress syndrome and passed away.

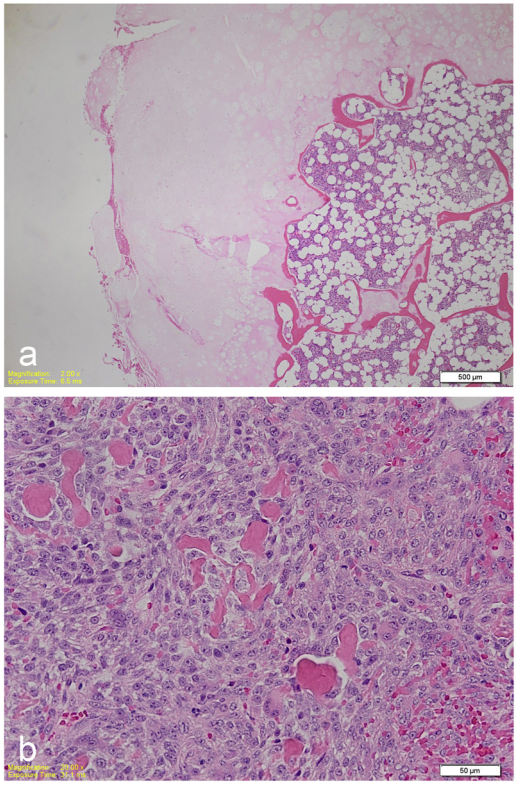

Fig. 2. (a) Osteochondroma: low power view of the cartilaginous cap overlying cancellous bone of the stalk. (b) Osteosarcoma: high power view of malignant tumour cells with intervening pink staining osteoid.

3. Discussion

Osteochondromas are the commonest benign tumour of bone. They are usually found in the metaphyseal region of the appendicular skeleton, with only 3% of solitary lesions occurring within the vertebral column. Multiple lesions are observed in patients with autosomal dominant condition hereditary exostoses. Approximately 50% of lesions occur in the cervical spine and involve the posterior elements of the vertebra, although any part may be involved [1]. They originate from within the periosteum and grow progressively by endochondral bone formation and are composed of cortical and medullary bone with an overlying hyaline cartilage cap. By comparison osteosarcomas are defined pathologically as primary intramedullary high grade malignant tumours in which the neoplastic cells produce osteoid in variable quantities. These lesions are also most commonly seen in metaphyseal regions with vertebral involvement being very rare representing 1–2% of all cases. They have a preponderance for the thoraco-lumbar spine and the vast majority are lytic in nature.

The presence of two distinct and rare tumour types within different regions of the spine in a patient with Gorlin's Syndrome, to the best of our knowledge, is hitherto undescribed. Both lesions were located in regions of the spine where they are rarely observed. Osteochrondroma typically affects the cervical spine, most often at the C2 level [1]. In the few reported cases involving the thoracic spine it has been observed that the lesions arise from the tip of the spinous or transverse process. The first observed lesion in our case was noted to arise from the spinous process of L2 with the subsequent lesion at T9 arising from the junction of pedicle and the vertebral body. Over two-thirds of primary osteosarcomas present within the thoracolumbar spine. Osteochondroma has previously