Diffuse scalp malignant peripheral nerve sheath tumor with intracranial extension in a patient with neurofibromatosis type 1

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ABSTRACT

We describe a rare scalp malignant peripheral nerve sheath tumor (MPNST) with cranial destruction and intracranial extension in a 52-year-old male with neurofibromatosis type 1 (NF1). The scalp tumor measured 22 cm × 18 cm, with local surface ulceration. Skin examination revealed many café-au-lait spots and small, hard dermal nodules on the trunk. CT scans revealed the scalp tumor to have heterogeneous density with partial destruction of the right parietal cranium; on T1-weighted MRI the scalp tumor displayed heterogeneous hypointensity, whereas on T2-weighted and fluid-attenuated inversion recovery MRI it was hyperintense. The tumor was excised totally and the scalp reconstructed using a skin flap isolated from the lateral aspect of the left thigh. Histological examination confirmed that the tumor was an MPNST. The patient recovered uneventfully and was well at the 6-month follow-up, with no local or other tumor recurrence noted.

1. Introduction

Malignant peripheral nerve sheath tumors (MPNST) are soft tissue sarcomas of ectomesenchymal origin, and are believed to be derived from the components of nerve sheaths such as perineural fibroblasts or Schwann cells. These unusual tumors are located mainly in the buttocks, thighs, brachial plexus and paraspinal regions. We report a male patient with a highly unusual MPNST in the scalp.

2. Case report

A 52-year-old man presented to our facility with a diffuse, multilobular, painless and non-mobile swelling on his head, which had been present for approximately eight-and-a-half years. During the first six years, the swelling was about the size of an egg and located in the right parietal region. Over the previous two years, it gradually increased in size, with an additional growth spurt during the past six months. The tumor was found to measure 22 cm × 18 cm and had local surface ulceration (Fig. 1A). Examination of his skin revealed numerous café-au-lait spots; on his trunk were small, hard dermal nodules the size of rice grains, which had been there since puberty. CT scans revealed the scalp tumor to have heterogeneous density with partial destruction of the right parietal cranium (Fig. 2A,B); on T1-weighted MRI the scalp tumor displayed heterogeneous hypointensity, whereas on T2-weighted and fluid-attenuated inversion recovery MRI it was hyperintense (Fig. 2C–E). During surgery, the extracranial swelling was noted isolated with at least a 2-cm margin of scalp on all borders; the destroyed cranium and associated intracranial extension were all excised and weighed approximately 1870 g (Fig. 1B,C). The scalp was repaired using a skin flap that was isolated from the lateral aspect of the left thigh (Fig. 1D). Six of the dermal nodules in the trunk were also excised for histological assessment. The histological features of the scalp swelling were consistent with those of an MPNST (Supplementary Fig. 1A–D), and the nodules from the trunk showed characteristics of neurofibromas. The patient recovered uneventfully and was well at the 6-month follow-up, with no localized or generalized tumor recurrence seen.

3. Discussion

MPNST occur in 0.001% of the general population, and can develop in pre-existing neurofibromas, *de novo* from peripheral nerve sheaths or following radiation therapy.[1,2] Using the criteria of the National Institutes of Health consensus, our patient was diagnosed as having neurofibromatosis type 1 (NF1), as the café-au-lait spots had existed since puberty and histological examination found the dermal nodules to be neurofibromas.[3] Although uncommon, MPNSTs have been reported to occur in many sites, however few instances of scalp MPNSTs have been previously reported. Of these, none mentioned a relationship with NF1.[2,4–6]

Although MPNSTs do not have specific imaging features, CT scans and MRI assessment are useful tools to detect the relationship with surrounding tissues.[7] Of the four previously reported patients with scalp MPNST, radiological assessment found three to have cranial destruction and intracranial extension; only one patient had neither cranial destruction nor intracranial extension.[5] Given that cranial destruction can be produced by other types of tumors, including a giant benign tumor, this is not a reliable indicator of MPNST. However, MPNSTs can increase in size rapidly over a short time, and this generally indicates malignant transformation[8]. Interestingly, MPNST are often located in the parietal or occipital scalp, and occasionally show associated ulceration and bleeding.[2,4,6] Histopathologically, MPNST are highly cellular tumors that have a fascicular pattern, spindle-shaped nuclei and scant cytoplasm. Immunoreactivity for the S100 protein suggests that the tumor originates from Schwann cells.[9,10] Our patient's tumor was histologically consistent with these features, and we concluded it originated from a Schwann cell. Microscopic demonstration of invasion into muscles and mitotic activity indicated malignancy. Some authors suggest that p16 can be used to differentiate MPNST from benign peripheral nerve sheath tumors (BPNST), as p16 is expressed in BPNST but not MPNST.[5] Agesen et al. felt that the gross rearrangements of the p16(INK4A) gene observed in most patients with MPNST explained the absence of the

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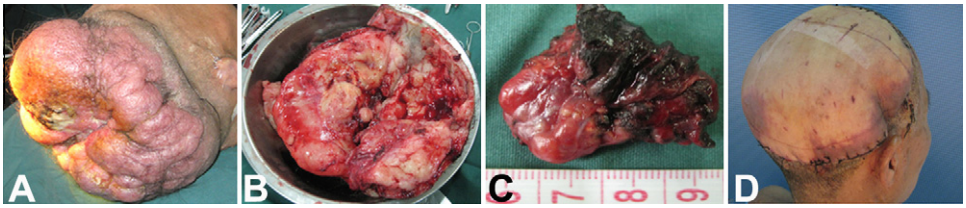

Fig. 1. Photographs of the tumor showing: (A) the pre-operative macroscopic image (B) the extracranial part of the tumor (C) the intracranial part of the tumor and (D) the appearance after the skin flap transplant. (This figure is available in colour at www.sciencedirect.com.)

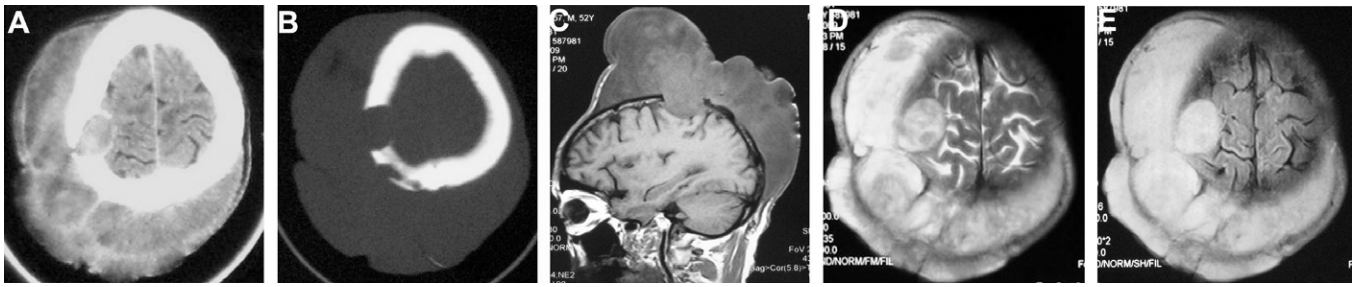

Fig. 2. Axial CT scans of the tumor showing: (A) heterogeneous density with (B) partial destruction of the right parietal cranium; and MRI of the tumor showing: (C) heterogeneous hypointensity on T1-weighted sagittal MRI, compared to (D) hyperintensity on T2-weighted axial and (E) fluid-attenuated inversion recovery axial MRI.

encoded protein of the p16 gene in these tumors.[11] Recently, Fang et al. examined of primary, metastatic and recurrent MPNSTs with underlying NF1 and found deletions of 9p, 12q21–q32 and complete loss of the X-chromosome, as well as gains in 17q25.[12]

Once the diagnosis of MPNSTs is suspected, surgery is the mainstay of treatment.[13] For our patient, the excised scalp margin was intra-operatively assessed repeatedly, to ensure total tumor excision (with clear margins). However, if the tumor is found to have involvement of important intracranial structures or blood vessels, partial resection in combination with radiotherapy may be a treatment alternative.[2]

Patients with MPNST have had a poor prognosis; for those who have NF1, the prognosis appears to be even worse. In the group reported by Stark et al., all patients experienced local tumor recurrence, and had a mean disease-free survival time of 10.6 months.[14] At his 6-month follow-up, our patient showed no sign of tumor recurrence; however, his recovery continues to be evaluated.

Appendix A. Supplementary material

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.jocn.2010.01.055.

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