On-line Case Report

Management of massive lower limb plexiform neurofibromatosis – when to intervene?

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ABSTRACT

Massive plexiform neurofibromatosis is an uncommon manifestation of Type I neurofibromatosis. These tumours result in functional disability and severe disfigurement. Due to the variable natural history of these lesions, the optimal management is not well defined and surgery is often delayed until significant disfigurement has occurred. The aim of this report is to discuss the optimum timing of surgical intervention and to highlight the difficulties in dealing with an advanced lesion.

Keywords: Neurofibromatosis I – Plexiform neurofibroma – Peripheral nervous system neoplasms

A 37-year-old man with type 1 neurofibromatosis presented for debulking of a massive plexiform neurofibroma of the left leg (Figs 1 and 2). The lesion presented at birth as an area of hyperpigmentation. There was underlying skeletal abnormality with limb discrepancy noted since infancy resulting in several fractures of the femur. The pigmented area remained unchanged until his late teens when it gradually increased in volume producing a disfiguring redundant soft-tissue mass. He had progressive bone involvement with pseudo-arthritis of the femur and unstable knee and hip joints such that weight-bearing was not possible in this limb.

Debulking surgery was carried out under tourniquet control. The tumour was very vascular containing thin-walled vessels and diathermy was not effective in controlling the bleeding (Fig. 3). The tourniquet was let down for brief intervals to identify bleeding points. Eventually, haemostasis was achieved by internal packing with gauze and partial closure. He required 10 units of

blood and the specimen excised weighed 11 kg. He was returned to theatre 2 days later for removal of packs and wound closure. Histological examination revealed plexiform neurofibromatosis with no evidence of malignancy. He subsequently underwent two further resections and, to date, there has been no significant recurrence in 2-year follow-up.

Discussion

Plexiform neurofibroma is reported to occur in 26.7% of patients with type I neurofibromatosis.¹ Plexiform neurofibromas present at, or soon after, birth as areas of hyperpigmentation, thickening of the skin and hair excess.² The optimal management of plexiform neurofibroma is not well defined and surgery is often delayed until significant disfigurement has occurred. The aim of this report is to discuss the optimum timing of surgical intervention and to highlight the difficulties in dealing with an advanced lesion.

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Figure 1 Massive left lower limb plexiform neurofibroma.

Figure 2 Magnetic resonance image showing diffusely infiltrating plexiform neurofibroma of the left thigh.

Figure 3 Very vascular tissue with large feeding vessels demonstrated.

Figure 4 Left leg 4 months after debulking surgery.

The natural history of these lesions is variable with some remaining superficial and asymptomatic throughout life and some progressing into large invasive disfiguring lesions.^{3,4} Friedrich *et al.*⁴ recently classified these lesions based on the magnetic resonance image (MRI) appearance – superficial, displacing and invasive. Superficial plexiform neurofibromas remain within the upper layer of the skin, usually not involving major nerves. The displacing type develops in deeper layers of the skin or within the body but does not invade adjacent muscles or skin. In contrast, invasive plexiform neurofibromas infiltrate multiple tissue planes to involve muscle and bone (Fig. 2). These are much more difficult or impossible to resect. It is not known if a superficial lesion can progress into the invasive type and it is not clear if early surgical intervention can halt or slow the progression of these tumours. Friedrich *et al.*⁵ recommended MR delineation and early excision of superficial lesions thus preventing possible progression. In a series of 9 children there was no recurrence in 4 years of follow-up.⁵

Resection and debulking of invasive plexiform neurofibromas is associated with a high rate of recurrence. In one paediatric series, complete resections developed recurrence in 20% and incomplete resections had a recurrence of up to 45%.⁶ A common theme is the vascularity of these lesions and their abnormal propensity to bleed (Fig. 3). Mukherji⁷ compared them to angiomas and also commented on the friability of the vessels. We were fortunate in this case that it was possible to use a tourniquet. Some authors recommend pre-operative angiography and embolisation prior to surgery, although experience with this is limited.⁸ In our experience, internal packing with gauze is an effective method of controlling the bleeding when normal measures fail. The life-long risk of malignant transformation for plexiform neurofibromas quoted in the more recent reviews is 2–5%.^{2,3} This may be associated with a rapid growth phase or pain of new onset. Such changes should prompt

urgent investigation with MRI scan and biopsy.

Due a lack of natural history data and the unpredictable growth patterns of plexiform neurofibromas, it remains difficult to advise about the best time to intervene surgically. It would appear that timely intervention could limit the disfigurement and morbidity associated with large lesions. There is some evidence that early excision of superficial lesions may prevent progression.^{5,6} One can reasonably speculate that earlier intervention in the case described would have made surgery less hazardous with less blood loss and easier flap design. There is a case for developing a national registry of such cases in specialist centres to facilitate patient monitoring and the development of appropriate treatment protocols.

References

1. Huson SM, Harper PS, Compston DA. Von Recklinghausen neurofibromatosis. A clinical and population study in south-east Wales. *Brain* 1988; **111**: 1355–81.
2. Gutmann DH, Aylsworth A, Carey JC, Korf B, Marks J, Pyeritz RE *et al.* The diagnostic evaluation and multidisciplinary management of neurofibromatosis 1 and neurofibromatosis 2. *JAMA* 1997; **278**: 51–7.
3. Woodruff JM. Pathology of tumours of the peripheral nerve sheath in type 1 neurofibromatosis. *Am J Med Genet* 1999; **89**: 23–30.
4. Friedrich RE, Korf B, Funsterer C, Mautner VF. Growth type of plexiform neurofibromas in NF1 determined on magnetic resonance images. *Anticancer Res* 2003; **23**: 949–52.
5. Friedrich RE, Schmelzle R, Hartmann M, Funsterer C, Mautner VF. Resection of small plexiform neurofibromas in neurofibromatosis type 1 children. *World J Surg Oncol* 2005; **3**: 6.
6. Needle MN, Cnaan A, Dattilo J, Phillips PC, Shochat S, Sutton LN *et al.* Prognostic signs in the surgical management of plexiform neurofibroma: the Children's Hospital of Philadelphia experience, 1974–1994. *J Pediatr* 1997; **131**: 678–82.
7. Mukherji MM. Giant neurofibroma of the head and neck. *Plast Recon Surg* 1974; **53**: 184–9.
8. Littlewood AHN, Stilwell JH. The vascular features of plexiform neurofibroma with some observations on the importance of pre-operative angiography and the value of pre-operative intra-arterial embolisation. *Br J Plast Surg* 1983; **36**: 501–6.