Plexiform Neurofibroma of the Thigh: Management Experience and Review of Literature

Echchaoui Abdelmoughit[1], Al Ahmad Ahmad[2], Benyachou Malika[1], Fathi Nahed[1], El Mazouz Samir[1], Gharib Nour-eddine[1], Abbassi Abdellah[1]

[1]*Department of Plastic and Reconstructive Surgery, Ibn Sina University Hospital, Rabat, Morocco*
[2]*Department of Orthopaedic Surgery & Traumatology, Ibn Sina University Hospital, Rabat, Morocco*

Conflict of Interest: None declared

This article has been peer reviewed.

Article Submitted on: 4th February 2015

Article Accepted on: 5th April 2015

Funding Sources: None declared

Correspondence to: Dr Echchaoui Abdelmoughit

Address: Department of Plastic and Reconstructive Surgery, Ibn Sina University Hospital, Rabat, Morocco

Email: e.moughit@hotmail.fr

Cite this Article: Abdelmoughit E, Ahmad AA, Malika B, Nahed F, Samir EM, Nour-eddine G, Abdellah A. Plexiform neurofibroma of the thigh: management experience and review of literature. J Pioneer Med Sci. 2015; 5(3):103-106

ABSTRACT

Plexiform neurofibromas (PNF) are benign nervous system tumors falling within the scope of neurofibromatosis type I, also known as von Recklinghausen's disease. PNF are currently treated surgically and surgery is required in many cases of symptomatic PNF. Surgery may result in added morbidity in some cases. Modelante surgery is proposed for giant and complex tumors, in which full excision is often impossible or mutilating. We report our management experience of a 20-year-old woman who presented with a large symptomatic PNF on her left thigh. It was causing difficulty in schooling and her social integration. The inextirpable nature of the lesion necessitated successive modelante resections which significantly improved her quality of life.

Keywords: Plexiform neurofibroma; Thigh; Modelante Surgery

INTRODUCTION

Plexiform neurofibromas (PNF) are benign peripheral nerve tumors that occur in neurofibromatosis type I (NF1). These tumors undergo progressive enlargement since childhood and over a period of time result in significant deformation of both aesthetic and functional importance [1].
Cases of lower extremity PNF are seen less frequently than those seen on the face and neck [2].
PNF treatment is currently surgical and must be carried out by experienced teams (plastic surgery, orthopedic surgery, and dermatology) [3]. Tumor resection is modelante, allowing for improved aesthetic and functional result, without requiring a full excision which is often impossible or too mutilating than the disease itself.

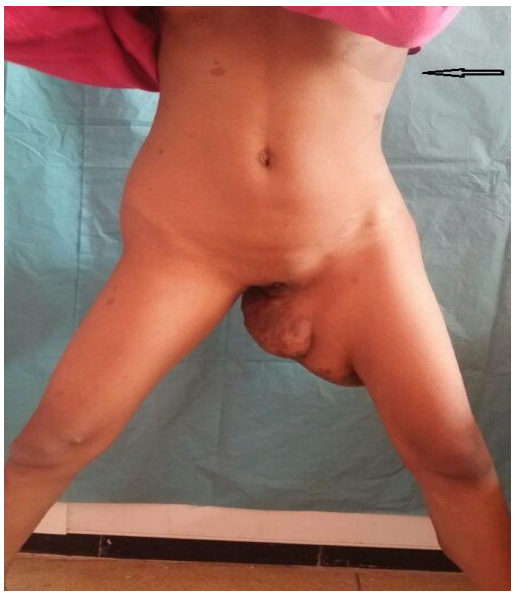
CASE REPORT

We report the case of a 20-year-old female with NF1 who presented with 4-months history of a growing mass on the left thigh causing difficulty in walking and in wearing trousers. She denied any history of local trauma or neurological symptoms, such as paresthesia or motor dysfunction. On physical examination, a large movable soft tissue mass was evident over the dorsal surface of her left thigh (Figures 1 and 2). The skin was supple without any discoloration. Muscle strength and vascular status were normal. She had several findings suggestive of a diagnosis of NF. For instance, multiple cafe-au-lait spots (Figure 2), axillary freckling, and a first-degree relative with NF1.
Plain-film radiographs were normal; magnetic resonance imaging (MRI) demonstrated a large soft tissue mass located on the posterior surface of thigh muscles. The mass appeared to infiltrate surrounding soft-tissue structures (sciatic nerve and its branches), making its margins difficult to delineate; the osseous structures were without lysis or erosion.
The inextirpable nature of the lesion obliged us to perform modelante resections under general anesthesia and subcutaneous infiltration with xylocaine containing adrenaline. Rigorous hemostasis was performed, and then skin closed over two suction drains (Figure 3).
The full lesion of approximately 25x8 cm was removed as three separate specimens. Histological examination confirmed the diagnosis of PNF and excluded malignancy. At one-year follow-up, there was a recurrence and the patient was scheduled again for a subtotal and palliative resection.

DISCUSSION

Figure 1: A large mass of PNF on the posterior surface of the left thigh which displaces the gluteal cleft

Figure 2: Frontal view showing a flat, ovoid, large, brown macule consistent with a café-au-lait spot (arrow)



Neurofibromas are benign peripheral nerve tumors that occur sporadically in patients with the autosomal dominant syndrome neurofibromatosis type 1 (NF1) [4]. NF1 is one of the most common genetic diseases, affecting approximately 1 in 3000 newborn infants [5, 6]. Plexiform neurofibromas (PNF) are neuro-fibroma variants in which tumor cells spread along multiple fascicles of the nerve, leading to a diffuse mass of thickened nerve fibers surrounded by a proteinaceous matrix [7]. Its morphologic appearance is classically compared to a "bag of worms" or "rosary beads" [8]. PNF are thought to be congenital and occur in 25% to 50% of children with NF1 [9]. The increased growth usually occurs in childhood and adolescence although there is some individual variation [1].

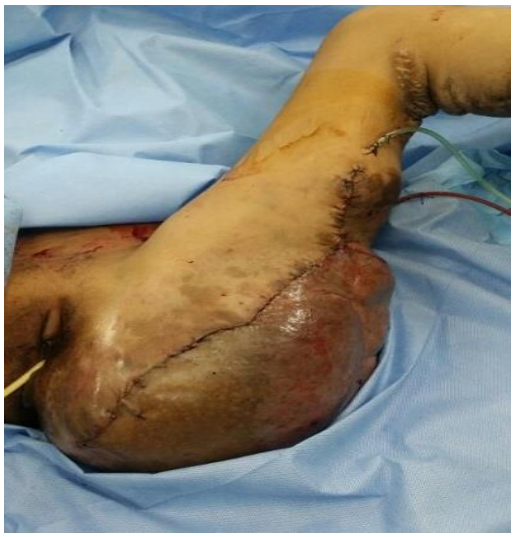
PNF may occur anywhere in the body, but in lower extremity are seen less frequently than those seen on the face and neck [2]. It develops as a nodular lesion confined to the nerve or a diffuse tumor that impinges on surrounding soft tissue and may be associated with bone hypertrophy, vascular change, overlying pigmentation, or excess hair growth [10].

Recently whole body MRI has facilitated the assessment of internal tumors in NF1, providing a useful tool for monitoring tumor burden and highlighting patients at increased risk for malignancy [11]. In our patient, MRI demonstrated a large soft tissue mass located on the posterior surface of the thigh muscles. The mass appeared to infiltrate the surrounding soft-tissue structures, making its margins difficult to delineate. There was no bone lysis or malignancy.

PNF can transform to malignant peripheral nerve sheath tumors (MPNSTs) with an estimated life time risk for patients with NF1 of approximately 10% [12-14]. So a careful clinical monitoring is warranted and should be adapted to each patient [15]. Furthermore, large PNF can compress vital organs and can result in severe morbidity and even death. Early diagnosis is important for genetic counseling and sometimes for therapeutic intervention [16].

Surgical excision of PNF is currently the only established therapy, but is rarely curative. Most of these tumors are invasive and not amenable to complete resection, as was seen in our case as well. Recent publication by Kolker et al [17] reported a case of a 55-year-old patient with a very large right thigh PNF removed successfully after preoperative embolization to reduce the bleeding risk at surgery. In our context, we performed a subcutaneous infiltration with xylocaine containing adrenaline to control hemostasis and to facilitate hydro-dissection. Surgical management of peripheral neurofibromas is controversial, with some authors recommending biopsy or excision, whereas others claim that malignant change in

Figure 3: Immediate postoperative appearance after modelante surgery



the tumor is possible as a result of operative trauma [18].
Hence, the risk versus the benefit of surgery must be weighed. Solitary neurofibromas often can be completely resected, with few complications [19, 20]. In some cases, however, total resection is simply not possible, because of the risk of neurologic injury, cosmetic disfigurement, or potential injury to adjacent vital anatomic structures.
A surgical team from the United States [21] has managed to successfully resect a giant PNF of the lower limb despite postoperative complications including lymphedema, cellulitis, and delayed wound closure, which were managed with antibiotics, ultrasound guided drainage, surgical revision of closure, compressive dressings, and vacuum-assisted wound closure. In our case, we couldn't perform a complete resection because of the inextirpable nature of the lesion and the risk of major post-operative complications.
This is especially true of PNF, which tend to be infiltrative and often envelop adjacent nerves, viscera, or vasculature. Indications for excision of solitary and PNF include lesions that cause significant symptomatic compression of critical anatomy, grow excessively, and cause significant pain as a result of neural compression [22]. Early detection and complete surgical excision are the only effective methods in the treatment of MPNSTs, because they are unresponsive to radiotherapy and chemotherapy, and have a tendency to metastasize [23, 24].
Additional therapeutic concerns involve patient and family education, pain management, adjunct-ive therapies, and psychological support. Patients and their families need to be completely aware of all aspects of the disease, including natural history, complications, and treatment options. Information concerning genetic counseling should be provided; pain is frequently managed with medications [3].

CONCLUSION

PNF of lower extremity are considered an uncommon skin tumor, they represent a challenge of surgical management in some locations if disfigurements, functional and aesthetic defects, and require a multidisciplinary approach.
The wide variability of clinical expressions, tumor risks and its totally unpredictable evolutionary nature requires regular long-term monitoring of patients with NF1.

REFERENCES

1. Tucker T, Friedman JM, Friedrich RE, Wenzel R, Fünsterer C, Mautner VF. Longitudinal study of neurofibromatosis 1 associated plexiform neurofibromas. *J Med Genet* 2009; 46:81–85.
2. Rekha A, Gopalan TR. Von Recklinghausen neurofibromatosis-pachydermatocele causing lower limb gigantism: a case report. *Int J Low Extrem Wounds* 2006; 5:61–63.
3. Gajeski BL, Kettner NW, Awwad EE, Boesch RJ. Neurofibromatosis Type I: clinical and imaging features of von Recklinghausen's disease. *J Manipulative Physiol Ther* 2003; 26:116-127.
4. Carroll SL, Ratner N. How does the Schwann cell lineage form tumors in NF1? *Glia* 2008; 56:1590-605.
5. Gutmann DH. The neurofibromatoses: when less is more. *Hum Mol Genet* 2001; 10:747-55.
6. Le LQ, Parada LF. Tumor microenvironment and neurofibromatosis type I: connecting the GAPs. *Oncogene* 2007; 26:4609-16.
7. Korf BR. Plexiform neurofibromas. *Am J Med Genet* 1999; 89:31-7.
8. Momoh JT. Plexiform neurofibroma in children. *East Afr Med J* 1986; 63:334-338.
9. Boulanger JM, Larbrisseau A. Neurofibromatosis type 1 in a pediatric population: Ste-Justine's experience. *Can J Neurol Sci* 2005; 32:225-31.
10. Ferner RE, Huson SM, Thomas N, Moss C, Willshaw H, Evans DG, et al. Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. *J Med Genet* 2007; 44:81–88.
11. Mautner VF, Asuagbor FA, Dombi E, Fünsterer C, Kluwe L, Wenzel R, et al. Assessment of benign tumor burden by whole-body MRI in patients with neurofibromatosis 1. *Neuro Oncol* 2008; 10:593-598.
12. Evans DG, Baser ME, McGaughran J, Sharif S, Howard E, Moran A. Malignant peripheral nerve sheath tumors in neurofibromatosis 1. *J Med Genet* 2002; 39:311-4.
13. Mc Caughan JA, Holloway SM, Davidson R, Lam WW. Further evidence of the increased risk for malignant peripheral nerve sheath tumours from a Scottish cohort of patients with neurofibromatosis type 1. *J Med Genet*

2007; 44:463-6.
14. Tucker T, Wolkenstein P, Revuz J, Zeller J, Friedman JM. Association between benign and malignant peripheral nerve sheath tumors in NF1. *Neurology* 2005; 65:205-11.
15. Pinson S, Wolkenstein P. Neurofibromatosis type 1 or Von Recklinghausen's disease. *Rev Med Int* 2005; 26:196–215.
16. Canale DJ, Bebin J. Von Recklinghausen's disease of the nervous system. In: Vinken PJ, Bruyn GW, eds. Handbook of Clinical Neurology. Volume 14. The phakomatoses. Amsterdam, New York: North-Holland Publishing Company. 1972: 132-62.
17. Kolker S, Wunder JS, Roche-Nagle G. Hybrid resection of a giant thigh plexiform neurofibroma. *Int J Surg Case Rep* 2015; 8:1-4.
18. Pollack IF, Mulvihill JJ. Neurofibromatosis 1 and 2. *Brain Pathol* 1997; 7:823-36.
19. Donner TR, Voorhies RM, Kline DG. Neural sheath tumors of major nerves. *J Neurosurg* 1994; 81:362-73.
20. Halliday Al, Sobel RA, Martuza RL. Benign spinal nerve sheath tumors: their occurrence sporadically and in neurofibromatosis types 1 and 2. *J Neurosurg* 1991; 74:248-53.
21. Ross AL, Panthaki Z, Levi AD. Surgical management of a giant plexiform neurofibroma of the lower extremity. *World Neurosurg* 2011; 75:754-7.
22. Pollack IF, Colak A, Fitz C, Wiener E, Moreland M, Mulvihill JJ. Surgical management of spinal cord compression from plexiform neurofibromas in patients with neurofibromatosis 1. *Neurosurgery* 1998; 43:248-55.
23. Korf BR. Malignancy in neurofibromatosis type 1. *Oncologist* 2000; 5:477-85.
24. Wong WW, Hirose T, Scheithauer BW, Schild SE, Gunderson LL. Malignant peripheral nerve sheath tumor: analysis of treatment outcome. *Int J Radiat Oncol Biol Phys* 1998; 42:351-60.