

## Case Report

# Isolated giant plexiform neurofibroma of lower back: a rare case presentation

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## ABSTRACT

Plexiform neurofibroma is a rare benign tumor of the peripheral tissue cells that develops in the perineurium that is often considered pathognomonic of neurofibromatosis type 1 (NF1 or von Recklinghausen disease). They occur most frequently in the craniomaxillofacial region, rarely on back and extremities. They are extremely vascular and there is 15-20% potential for malignant transformation. A 35-year-old married female presented with painless swelling in left lower back that began at around 8 years of age. The swelling gradually increased in size. The patient reported embarrassment over the disfigurement caused by the mass. Her medical history was unremarkable and none of the relatives was known to be affected. Surgical resection of the swelling with primary closure was done. Histopathology findings were consistent with neurofibromatosis. Hereby reporting a rare case of isolated plexiform neurofibroma of lower back which was surgically cured as a perusal of rare entity. We also try to emphasize on the need of sprightly clinical diagnosis with multidisciplinary approach in the management of these type of tumors. Finally, we insist on the need of a long term clinical and radiological follow-up of these patients to assess post resection recurrence or malignant transformation.

**Keywords:** Giant plexiform neurofibroma, Back, Neurofibromatosis type 1

## INTRODUCTION

Plexiform neurofibroma (PN) is a rare benign tumor of the peripheral tissue cells that develops in the perineurium that is often considered pathognomonic of neurofibromatosis type 1 (NF1 or von Recklinghausen disease).<sup>1</sup> There are three main types of neurofibromas: localized (most common), diffuse, and plexiform. Although the majority of neurofibromas occur sporadically and have an extremely low risk of malignant transformation, the plexiform type is pathognomonic for NF 1 and carries an increased risk of malignant transformation.<sup>2</sup> It is important to stress that “plexiform” does not imply involvement of a nerve plexus, such as the lumbar or brachial plexus, although these sites can be involved. Rather, the term implies a network-like growth

of neurofibroma involving multiple fascicles of a nerve and may include multiple branches of a large nerve; leading to a diffuse mass of thickened nerves.<sup>3</sup> Surgery is the main stay for treatment. Surgery is frequently reserved until PNs have progressed to the point of causing functional compromise, aesthetic deformity or pain.<sup>4</sup>

## CASE REPORT

A 35-year-old married female presented with painless swelling in left lower back that began at late childhood (at around 8 years of age). The swelling gradually increased in size. The patient reported embarrassment over the disfigurement caused by the mass with no neurological complaints. Skin examination revealed no

cutaneous neurofibromas and cafe - au - lait macules on the trunk and arms. However, skin excoriations found at two sites of 2×1 cm over the swelling. Her blood pressure was within normal limits. Her medical history was unremarkable and none of the relatives was known to be affected. No other stigma of neurofibromatosis syndrome were identified. Routine laboratory tests were normal. Ophthalmological examination was normal. Magnetic resonance imaging (MRI) brain was done to rule out any focal lesions in brain. It revealed no significant abnormality. No signal intensity noted in adjacent bone. On MRI spine, an ill-defined focal lesion was seen in subcutaneous plane extending from L1 to S1 vertebral body level with large exophytic component extending till the gluteal region. It was iso to hypointense to muscle on T1 and T2 and showed mild homogenous post contrast enhancement. Decision was taken to go with complete resection of the swelling.



**Figure 1: Dorsal view.**



**Figure 2: Lateral view.**

Following the tumor removal primary skin closure was achieved with romovac suction drain insitu. Postoperative course was uneventful and drain was removed on post op day 4 and patient discharged. The gross specimen measured 31×23×4.5 cm and weighed 2.1 kg. Skin surface of the swelling appeared hyperpigmented with hypopigmentation near its root. Also, skin surface appeared wrinkled with multinodularity felt on palpation. On Histopathology, tumor mass was composed of neurofibromatous lobules (multiple fascicles) with central nerve bundles. Myxomatous change was seen in stroma. Tumor cells were spindle shaped with slender wavy elongated nuclei and ill-defined cytoplasmic border.

Increased vascularity was seen in many areas. Tumor was present in deeper dermis and extending into subcutaneous adipose tissue. Malignant transformation was not seen in the tumor. On follow-up the wound healed well, sutures were removed on post op day 14 and resulted in a normal calibre and functioning limb.



**Figure 3: Neurofibroma hanging on left side of trunk, show the upper margin and extent of neurofibroma, neurofibroma is seen crossing the midline.**



**Figure 4: Intra operative picture after resection of swelling and before closure of wound with romovac suction drain in situ.**



**Figure 5: Post resection specimen.**

## DISCUSSION

A neurofibroma is a benign nerve sheath tumor consisting of fibroblasts, Schwann cells, and neural elements that expand and diffusely infiltrate the nerve. Neurofibromas have been subdivided into two broad categories: dermal and plexiform. Dermal neurofibromas are associated with a single peripheral nerve, while PNs are associated with multiple nerve bundles forming interdigitating network of finger-like fronds of tumor.<sup>5</sup>

**Table 1: National Institutes of Health diagnostics criteria for NF1.**

Two or more of the following clinical features must be present
Six or more café-au-lait macules (>5 mm in greatest diameter in prepubertal individuals or <15 mm greatest in postpubertal individuals)
Two or more neurofibromas of any type or one PN
Freckling in the axillary or inguinal regions
Optic glioma
Two or more iris hamartoma (Lisch nodules)
Distinctive bony lesion such as sphenoid dysplasia, or thickening of the long bone cortex with or without pseudoarthrosis
A first-degree relative (parent, sibling or offspring) with NF 1 based on the above criteria.

Neurofibromatosis appears in two different neurocutaneous autosomal dominant genetic forms. NF types 1 and 2 affect nerve cell tissue development and growth.<sup>6</sup> NF1, also known as von Recklinghausen's disease is the most common type of Neurofibromatosis (Table 1). PNs occur in 26.7% of patients with NF1.<sup>7</sup> They can be present at birth, but often appear between the ages of 2 and 5.<sup>8</sup> It has also been described as an isolated entity. It is frequently located on the head and neck due to the rich innervations of the area; however, they have also been described in the extremities, where they follow the axis of a nervous track, appearing in a cord-like form.<sup>9,10</sup> This patient presented with a isolated giant diffuse PN on lower back without NF1 that had been growing for more than 20 years. There is a significant discrepancy in the size of tumors that are described as "giant" neurofibromas. Some authors reserve this term for tumors weighing more than 20 kg, even though no clear consensus has been reached.<sup>11</sup> The hallmark of NF1 is the hyperpigmented cutaneous lesions. Diagnostic criteria were developed by a National Institutes of Health consensus conference in 1987 and refined by a second conference in 1997 (Table 1).<sup>12</sup>

In order to investigate the neurofibroma MRI and computed tomography are used to determine the site and the expansion of the plexiform neurofibroma. Unfortunately, however, these imaging modalities are not reliable in deciphering between a PN and those that have progressed into malignant peripheral nerve sheath

tumors.<sup>13</sup> In these patients surgery is not only a therapeutic but also an important diagnostic option. PN is usually a clinical diagnosis, however, the gold standard in identifying malignant transformation within a PN is histology. Unfortunately histology requires complete resection, which is not always possible. If a core biopsy is performed, the malignant change may be missed within a large heterogeneous tumor.<sup>14</sup> Pain, aesthetic issues and neurological involvements are known potential complications of plexiform neurofibromatosis over the certain areas of body, and in a series. Malignant transformation is the leading complication of plexiform neurofibroma. The lifetime risk of this transformation is estimated at 15 to 20% in patients with NF-1(4), most commonly presenting during adulthood at approximately 20 to 50 years old. Findings which favour malignant over benign peripheral nerve sheath tumors include: enlarging mass, size greater than 5 cm, ill-defined margins, perilesional edema, peripheral enhancement pattern, lack of central hypo intense "target" on T2 sequences, and heterogeneity with central necrosis.<sup>15</sup> These signs must force us to realize a radiological examination (MRI) and consider an early surgical revision.<sup>16</sup>



**Figure 6: (A) Postoperative day 14, sutures removed. Healthy wound (lateral view), (B) patient in prone position.**

With regard to the treatment of these giant tumors, surgical treatment still remains the mainstream, aiming at improving functional and aesthetic effect.<sup>17-19</sup> There is a conflict amongst literature about the right time and extent



of PN resection. Some literature suggests that removal of PN in early stage can restricts the extent of local involvement. But on the contrary Gutmann et al, stated that since complete removal is not feasible sometimes, so in asymptomatic children resection is not justified due to chances of significant regrowth potential of remnant tumor.<sup>20,21</sup> However, it is agreed that the most difficult challenge for this surgery is the variation of anatomy and unclear tumor margin. In addition, the tumor contains thickened cord of nerves along with abundant abnormal vessels; therefore, excessive hemorrhage during the operation is also a brainteaser. Furthermore, if the nerve roots of neurofibromas are from major nerves, surgical resection may result in functional impairment. Therefore, the surgeon must cautiously design a detailed preoperative treatment strategy.<sup>22</sup> Sometimes, if surgical management is used, the lesion's vascularity and its abnormal propensity to bleed must be acknowledged. Some have even compared PN lesions to angiomas as well as highlighted the friable nature of the lesion's vessels.<sup>23</sup> For this reason it has been recommended to use pre-operative angiography and embolization prior to surgery.<sup>24</sup> No prior embolization was performed in our case.

A complete resection is often difficult due to the extensive growths of the tumors and invasion of the surrounding tissues, beneficent bleeding and fragile tendency and therefore re-growth after surgery is common. High recurrence tendency of PN is more with its invasive subtype. In comparison to complete removal (20%), incomplete resection of PN has (44%) more chances of recurrence.<sup>25</sup> Largest series of surgically managed PN by Needle et al, (in 10 year of follow up) found 54% recurrence rate, and interestingly it reoccurred more in the head and neck area.<sup>26</sup>

For the giant neurofibroma of the back of in our patient, resection of the tumor was successfully performed and the defect was primarily closed. Postoperative wound infection and skin necrosis was not observed. Patient had excellent recovery after resection and no recurrence after one year of follow up till date.

## CONCLUSION

Hereby reporting a rare case of isolated PN of lower back which was surgically cured as a perusal of rare entity. We also try to emphasize on the need of sprightful clinical diagnosis with multidisciplinary approach in the management of these type of tumors. Finally, we insist on the need of a long term clinical and radiological follow-up of these patients to assess post resection recurrence or malignant transformation.

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